Targeted sequencing for liquid biopsy cancer research

Growing menu of assays provides new content for more relevant insights

Ion Torrent™ Oncomine™ Cell-Free Nucleic Acid (cfNA) Research Assays are highly sensitive, multibiomarker next-generation sequencing (NGS) assays that enable molecular insights into tumor evolution and detection of primary driver and resistance mutations. The high-value gene content includes key mutations identified by the OncoNetwork consortium and other clinical researchers around the world.

Based on multiplex PCR, our proprietary technology together with Ion Torrent™ sequencing enables researchers to develop tests that may impact treatment selection, treatment monitoring, and recurrence monitoring in the future.

Key benefits

- **Streamlined workflow**—complete NGS research workflow, from blood sample to variant data in just 2–3 days

- **Optimized content**—SNVs, indels, CNVs, and fusions for multibiomarker analysis

- **Flexible input amounts**—results enabled from one tube of blood

- **Low limit of detection**—variant detection down to 0.1% from targeted NGS assays

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The family of Oncomine cfNA Research Assays

Oncomine cfNA Research Assays enable research studies on tumor heterogeneity and recurrence, from minimal sample input (Figure 1). The assays achieve high correlation between variants called in formalin-fixed, paraffin-embedded (FFPE) samples and in cell-free DNA (cfDNA) from plasma (Table 1).
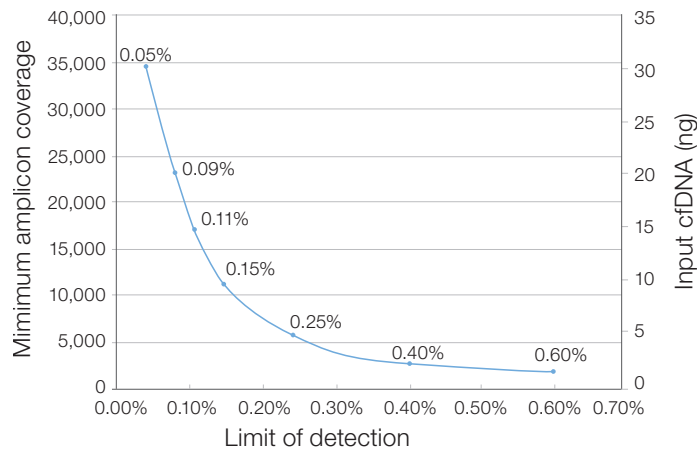
Built on proprietary, amplification-based technology, Oncomine cfNA Research Assays enable detection of driver and resistance mutations from cell-free nucleic acids, down to 0.1%. Select from five focused, tumor type–specific assays predesigned with key gene content, or a broad pan-cancer assay that covers all classes of mutations across multiple cancer types (Figure 2).

Figure 1. Amplicon coverage and input material determine limit of detection using Oncomine cfNA Research Assays.

Table 1. Correlation between results from FFPE and matched plasma samples (late-stage lung cancer samples).

Sample	Variant	FFPE samples	cfDNA
1	*EGFR*-L858R	**71.42%**	**2.62%**
2	*TP53*-R158L	**51.89%**	**4.32%**
3	*MET*-T1010I	43.87%	51.75%
	KRAS-G12C	**34.62%**	**0.28%**
4	NA	No detection	No detection
5	*EGFR*-L858R	**58.44%**	**7.28%**
	MET-T1010I	41.93%	48.72%
	TP53-Y220C	**35.54%**	**1.93%**
6	*TP53*-R158L	**10.19%**	**1.26%**

Values in boldface indicate somatic mutations; values not in boldface indicate germline mutations. As expected, there is a higher fraction of somatic mutations in FFPE samples than in plasma samples. Germline variants are seen at the expected levels of ~50% in both sample types. Data were obtained using the Oncomine Lung cfDNA Assay.

🫁 Lung		👤 Breast		🧬 Colon
Oncomine Lung cfDNA Assay	**Oncomine Lung Cell-Free Total Nucleic Acid Assay**	**Oncomine Breast cfDNA Assay**	**Oncomine Breast cfDNA Assay v2**	**Oncomine Colon cfDNA Assay**
ALK *MET* *BRAF* *NRAS* *EGFR* *PIK3CA* *ERBB2* *ROS1* *KRAS* *TP53* *MAP2K1*	*ALK* *MET* *BRAF* *NRAS* *EGFR* *PIK3CA* *ERBB2* **RET** *KRAS* *ROS1* *MAP2K1* *TP53*	*AKT1* *FBXW7* *EGFR* *KRAS* *ERBB2* *PIK3CA* *ERBB3* *SF3B1* *ESR1* *TP53*	*AKT1* *FBXW7* **CCND1** **FGFR1** *EGFR* *KRAS* *ERBB2* *PIK3CA* *ERBB3* *SF3B1* *ESR1* *TP53*	*AKT1* *ERBB2* *NRAS* *APC* *FBXW7* *PIK3CA* *BRAF* *GNAS* *SMAD4* *CTNNB1* *KRAS* *TP53* *EGFR* *MAP2K1*
• 11 genes • DNA only • 35 amplicons • 169 hotspots and indels	• 12 genes • Single library from DNA and RNA • 58 amplicons • >169 hotspots and indels • 49 fusions: *ALK, RET, ROS1* • CNV: *MET* • *MET* exon 14 skipping (3)	• 10 genes • DNA only • 26 amplicons • 152 hotspots and indels	• 12 genes • Single library to detect SNVs and CNVs • 76 amplicons • >152 hotspots and indels • CNVs: *CCND1, ERBB2, FGFR1* • More complete coverage of *TP53*	• 14 genes • DNA only • 49 amplicons • 236 hotspots and indels

⚛ Pan-cancer			
Oncomine Pan-Cancer Cell-Free Assay			
Hotspot genes	**Tumor suppressor genes**	**CNV genes**	**Gene fusions**
AKT1 *EGFR* *FLT3* *KRAS* *PDGFRA* *ALK* *ERBB2* *GNA11* *MAP2K1* *PIK3CA* *AR* *ERBB3* *GNAQ* *MAP2K2* *RAF1* *ARAF* *ESR1* *GNAS* *MET* *RET* *BRAF* *FGFR1* *HRAS* *MTOR* *ROS1* *CHEK2* *FGFR2* *IDH1* *NRAS* *SF3B1* *CTNNB1* *FGFR3* *IDH2* *NTRK1* *SMAD4* *DDR2* *FGFR4* *KIT* *NTRK3* *SMO*	*APC* *FBXW7* *PTEN* *TP53*	*CCND1* *ERBB2* *CCND2* *FGFR1* *CCND3* *FGFR2* *CDK4* *FGFR3* *CDK6* *MET* *EGFR* *MYC*	*ALK* *FGFR3* *BRAF* *MET* *ERG* *NTRK1* *ETV1* *NTRK3* *FGFR1* *RET* *FGFR2* *ROS1*
• 52 genes • Single library from DNA and RNA	• 272 amplicons • >900 hotspots and indels	• Extended coverage of *TP53* • 96 fusions	• 12 CNVs • *MET* exon 14 skipping

Figure 2. Gene content of Oncomine cfNA Research Assays. Additional genes included in new assays are in boldface.

Streamline your targeted sequencing workflow

The Oncomine cfNA workflow consists of three key steps (Figure 3). During sample preparation, cell-free nucleic acids are extracted, enriched, and amplified. These amplicon-based libraries are then assembled overnight before targeted resequencing. Our integrated informatics solution then takes you from variant caller to a finished report that provides contextual insight for sample-specific variants and their use with respect to labels, guidelines, and current global clinical trials (Figure 4). This process transforms data into knowledge, helping you gain efficiency for cancer research and future drug development.

"What's in the blood is actually what's relevant. One could argue it's not the overall tumor composition that we really want. We want the biologically relevant population. How do we define that? If that's in the blood, that's what we should be sampling— it's that global representation of tumor biology that we need."

—Minetta Liu, MD
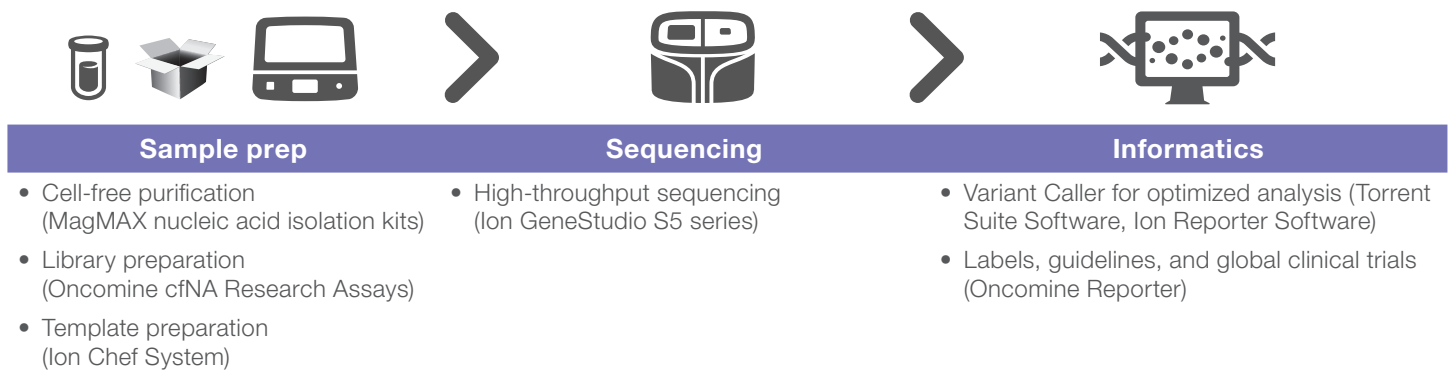Associate Professor of Oncology,
Mayo Clinic

Sample prep	Sequencing	Informatics
• Cell-free purification (MagMAX nucleic acid isolation kits) • Library preparation (Oncomine cfNA Research Assays) • Template preparation (Ion Chef System)	• High-throughput sequencing (Ion GeneStudio S5 series)	• Variant Caller for optimized analysis (Torrent Suite Software, Ion Reporter Software) • Labels, guidelines, and global clinical trials (Oncomine Reporter)

Figure 3. A 2–3 day comprehensive NGS workflow for liquid biopsy provides streamlined detection and analysis of genes and key mutations.

"Genomic heterogeneity after anti-EGFR therapy was successfully detected in 94% of the metastatic colorectal cancer research samples using a next-generation sequencing multibiomarker approach. The Oncomine Colon cfDNA Assay enabled detection of somatic mutations with MAF >0.1%."

–Beatriz Bellosillo, PhD
Hospital del Mar, Spain

Figure 4. Variant Caller plug-in for Ion Reporter™ Software.

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Ordering information

Product	Description	Cat. No.
Sample preparation		
MagMAX Cell-Free Total Nucleic Acid Isolation Kit	Isolation and enrichment kit for cell-free total nucleic acid (DNA and RNA)	A36716
MagMAX Cell-Free DNA Isolation Kit	Isolation and enrichment kit for cfDNA	A29319
Library preparation		
Oncomine Pan-Cancer Cell-Free Assay	Amplicon-based assay for targeted resequencing of cell-free DNA and RNA from pan-cancer research samples; includes hotspots, CNVs, fusions, and tumor suppressor genes	A37664
Oncomine Lung Cell-Free Total Nucleic Acid Assay	Amplicon-based assay for targeted resequencing of cell-free DNA and RNA from lung cancer research samples; includes >150 hotspots, CNVs, fusions, and *MET* exon 14 skipping	A35864
Oncomine Lung cfDNA Assay	Amplicon-based assay for targeted resequencing of cfDNA from lung cancer research samples; includes >150 hotspots	A31149
Oncomine Breast cfDNA Assay v2	Amplicon-based assay for targeted resequencing of cfDNA from breast cancer research samples; includes hotspots, CNVs, and expanded coverage of *TP53*	A35865
Oncomine Breast cfDNA Assay	Amplicon-based assay for targeted resequencing of cfDNA from breast cancer research samples; includes >150 hotspots	A31183
Oncomine Colon cfDNA Assay	Amplicon-based assay for targeted resequencing of cfDNA from colon cancer research samples; includes >150 hotspots	A31182
Tag Sequencing Barcode Set 1-24	Set of 24 unique barcode adapters, enabling multiplexing of amplicon library samples	A31830
Tag Sequencing Barcode Set 25-48		A31847
Template preparation		
Ion Chef System	Automates template preparation and Ion AmpliSeq library preparation	4484177
Ion 550 Kit-Chef	Prepackaged template and sequencing reagent cartridges with integrated sample tracking; for use with Ion 550 Chip Kit	A34541
Ion 540 Kit-Chef	Prepackaged template and sequencing reagent cartridges with integrated sample tracking; for use with Ion 540 Chip Kit	A30011
Ion 510 & Ion 520 & Ion 530 Kit-Chef	Prepackaged template and sequencing reagent cartridges with integrated sample tracking; for use with Ion 530 Chip Kit	A34461
Next-generation sequencing		
Ion GeneStudio S5 Prime System		A38196
Ion GeneStudio S5 Plus System	Next-generation sequencing instrument	A38195
Ion GeneStudio S5 System		A38194
Ion 550 Chip Kit	8 barcoded chips for sample tracking and sequencing, generating 100–130 million reads	A34538
Ion 540 Chip Kit	8 barcoded chips for sample tracking and sequencing, generating 60–80 million reads	A27766
Ion 530 Chip Kit	8 barcoded chips for sample tracking and sequencing, generating 15–20 million reads	A27764
Variant Caller and data analytics		
Ion Reporter Server System	Includes first-year software license and 3-hour online training	4487118
Oncomine Reporter	Software tool that creates custom reports and links variants to labels, guidelines, and global clinical trials	A34298

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

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