

The family of OncoPrint liquid biopsy assays

OncoPrint liquid biopsy 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).

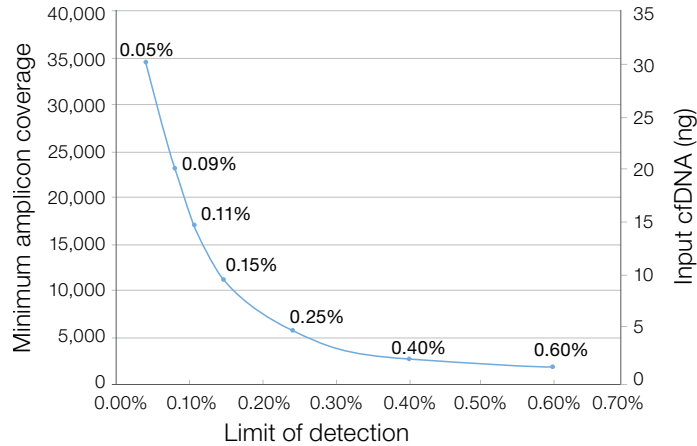


Figure 1. Amplicon coverage and input material determine limit of detection using OncoPrint liquid biopsy assays.

Built on proprietary, amplification-based technology, OncoPrint liquid biopsy 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 two broad pan-cancer assays that cover all classes of mutations across multiple cancer types (Figure 2).

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

Sample	Variant	FFPE	Plasma
1	<i>EGFR</i> -L858R	71.42%	2.62%
2	<i>TP53</i> -R158L	51.89%	4.32%
3	<i>MET</i> -T1010I	43.87%	51.75%
	<i>KRAS</i> -G12C	34.62%	0.28%
4	NA	No detection	No detection
	<i>EGFR</i> -L858R	58.44%	7.28%
5	<i>MET</i> -T1010I	41.93%	48.72%
	<i>TP53</i> -Y220C	35.54%	1.93%
6	<i>TP53</i> -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 OncoPrint Lung cfDNA Assay.

Lung				Breast				Colon		
OncoPrint Lung cfDNA Assay		OncoPrint Lung Cell-Free Total Nucleic Acid Assay		OncoPrint Breast cfDNA Assay		OncoPrint Breast cfDNA Assay v2		OncoPrint Colon cfDNA Assay		
<i>ALK</i>	<i>MET</i>	<i>ALK</i>	<i>MET</i>	<i>AKT1</i>	<i>FBXW7</i>	<i>AKT1</i>	<i>FBXW7</i>	<i>AKT1</i>	<i>ERBB2</i>	<i>NRAS</i>
<i>BRAF</i>	<i>NRAS</i>	<i>BRAF</i>	<i>NRAS</i>	<i>EGFR</i>	<i>KRAS</i>	<i>CCND1</i>	<i>FGFR1</i>	<i>APC</i>	<i>FBXW7</i>	<i>PIK3CA</i>
<i>EGFR</i>	<i>PIK3CA</i>	<i>EGFR</i>	<i>PIK3CA</i>	<i>ERBB2</i>	<i>PIK3CA</i>	<i>EGFR</i>	<i>KRAS</i>	<i>BRAF</i>	<i>GNAS</i>	<i>SMAD4</i>
<i>ERBB2</i>	<i>ROS1</i>	<i>ERBB2</i>	<i>RET</i>	<i>ERBB3</i>	<i>SF3B1</i>	<i>ERBB2</i>	<i>PIK3CA</i>	<i>CTNNB1</i>	<i>KRAS</i>	<i>TP53</i>
<i>KRAS</i>	<i>TP53</i>	<i>KRAS</i>	<i>ROS1</i>	<i>ESR1</i>	<i>TP53</i>	<i>ERBB3</i>	<i>SF3B1</i>	<i>EGFR</i>	<i>MAP2K1</i>	
<i>MAP2K1</i>		<i>MAP2K1</i>	<i>TP53</i>			<i>ESR1</i>	<i>TP53</i>			

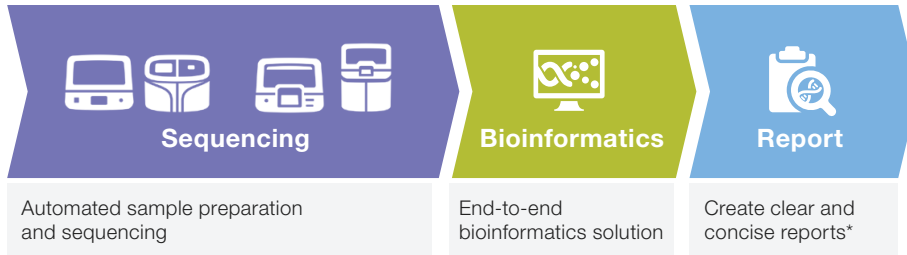
Pan-cancer										
OncoPrint Pan-Cancer Cell-Free Assay										
DNA hotspots					Tumor suppressors			CNVs		Fusions
<i>AKT1</i>	<i>EGFR</i>	<i>FLT3</i>	<i>KRAS</i>	<i>PDGFRA</i>	<i>APC</i>		<i>CCND1</i>	<i>ERBB2</i>	<i>ALK</i>	<i>FGFR3</i>
<i>ALK</i>	<i>ERBB2</i>	<i>GNA11</i>	<i>MAP2K1</i>	<i>PIK3CA</i>	<i>FBXW7</i>		<i>CCND2</i>	<i>FGFR1</i>	<i>BRAF</i>	<i>MET</i>
<i>AR</i>	<i>ERBB3</i>	<i>GNAQ</i>	<i>MAP2K2</i>	<i>RAF1</i>	<i>PTEN</i>		<i>CCND3</i>	<i>FGFR2</i>	<i>ERG</i>	<i>NTRK1</i>
<i>ARAF</i>	<i>ESR1</i>	<i>GNAS</i>	<i>MET</i>	<i>RET</i>	<i>TP53</i>		<i>CDK4</i>	<i>FGFR3</i>	<i>ETV1</i>	<i>NTRK3</i>
<i>BRAF</i>	<i>FGFR1</i>	<i>HRAS</i>	<i>MTOR</i>	<i>ROS1</i>			<i>CDK6</i>	<i>MET</i>	<i>FGFR1</i>	<i>RET</i>
<i>CHEK2</i>	<i>FGFR2</i>	<i>IDH1</i>	<i>NRAS</i>	<i>SF3B1</i>			<i>EGFR</i>	<i>MYC</i>	<i>FGFR2</i>	<i>ROS1</i>
<i>CTNNB1</i>	<i>FGFR3</i>	<i>IDH2</i>	<i>NTRK1</i>	<i>SMAD4</i>						
<i>DDR2</i>	<i>FGFR4</i>	<i>KIT</i>	<i>NTRK3</i>	<i>SMO</i>						

OncoPrint Precision Assay GX										
DNA hotspots					CNVs			Fusions		
<i>AKT1</i>	<i>CDKN2A</i>	<i>FGFR1</i>	<i>HRAS</i>	<i>MTOR</i>	<i>RAF1</i>	<i>ALK</i>	<i>FGFR1</i>	<i>ALK</i>	<i>FGFR3</i>	<i>RET</i>
<i>AKT2</i>	<i>CHEK2</i>	<i>FGFR2</i>	<i>IDH1</i>	<i>NRAS</i>	<i>RET</i>	<i>AR</i>	<i>FGFR2</i>	<i>AR</i>	<i>MET</i>	<i>ROS1</i>
<i>AKT3</i>	<i>CTNNB1</i>	<i>FGFR3</i>	<i>IDH2</i>	<i>NTRK1</i>	<i>ROS1</i>	<i>CD274</i>	<i>FGFR3</i>	<i>BRAF</i>	<i>NRG1</i>	<i>RSPO2</i>
<i>ALK</i>	<i>EGFR</i>	<i>FGFR4</i>	<i>KIT</i>	<i>NTRK2</i>	<i>SMO</i>	<i>CDKN2A</i>	<i>KRAS</i>	<i>EGFR</i>	<i>NTRK1</i>	<i>RSPO3</i>
<i>AR</i>	<i>ERBB2</i>	<i>FLT3</i>	<i>KRAS</i>	<i>NTRK3</i>	<i>TP53</i>	<i>EGFR</i>	<i>MET</i>	<i>ESR1</i>	<i>NTRK2</i>	
<i>ARAF</i>	<i>ERBB3</i>	<i>GNA11</i>	<i>MAP2K1</i>	<i>PDGFRA</i>		<i>ERBB2</i>	<i>PIK3CA</i>	<i>FGFR1</i>	<i>NTRK3</i>	
<i>BRAF</i>	<i>ERBB4</i>	<i>GNAQ</i>	<i>MAP2K2</i>	<i>PIK3CA</i>		<i>ERBB3</i>	<i>PTEN</i>	<i>FGFR2</i>	<i>NUTM1</i>	
<i>CDK4</i>	<i>ESR1</i>	<i>GNAS</i>	<i>MET</i>	<i>PTEN</i>						

Figure 2. Gene content of OncoPrint liquid biopsy assays. (A) Gene lists for five assays that target breast, colon, and lung cancer. (B) Gene lists for two pan-cancer assays, OncoPrint Pan-Cancer Cell-Free Assay and OncoPrint Precision Assay GX.

Streamline your targeted sequencing workflow

The Oncomine liquid biopsy NGS workflow consists of three key steps (Figure 3). First, cell-free nucleic acids are extracted, enriched, and amplified from one tube of blood. These amplicon-based libraries are then assembled and prepped for targeted resequencing. Our integrated informatics solution then takes you from variant caller to a clear and concise report that links biomarkers to relevant evidence (Figure 4). This process transforms data into knowledge, helping you gain efficiency for cancer research and future drug development.



* Specimen-to-report workflow will be available after the Ion Torrent™ Genexus™ Purification System and integrated reporting capabilities are added in 2021. The content herein may relate to products or workflows that have not been officially released or fully validated and is subject to change without notice.

Figure 3. Our 1- to 3-day comprehensive NGS workflow for liquid biopsy provides streamlined detection and analysis of genes and key mutations.

“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

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Sample ID: 00-123456789 Date: 29 Sep 2020 1 of 74

Sample Type: Blood Primary Tumor Site: Lung
Sample ID: 5555 Sample Collected: 010101

Sample Cancer Type: Non-Small Cell Lung Cancer

Relevant Non-Small Cell Lung Cancer Findings

Gene	Finding	Gene	Finding
ALK	Not detected	NTRK1	Not detected
BRAF	Not detected	NTRK2	Not detected
EGFR	EGFR E746_A750del (EGFR exon 19 deletion), EGFR T790M	NTRK3	Not detected
ERBB2	Not detected	RET	Not detected
KRAS	Not detected	ROS1	Not detected
MET	Not detected		

Relevant Biomarkers

Tier	Genomic Alteration	Annotations
IA	EGFR E746_A750del EGFR exon 19 deletion epidermal growth factor receptor Locus: chr7:55243484 Transcript: NM_005228.4
IA	EGFR T790M epidermal growth factor receptor Locus: chr7:55249771 Transcript: NM_005228.4
IIC	TP53 P60fs tumor protein p53 Locus: chr17:7579508 Transcript: NM_005545.5

Figure 4. Sample report from Ion Torrent™ Oncomine™ Reporter. The final report will provide relevant and detailed biomarker insights for your sample(s).

“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

Ordering information

Product	Cat. No.
Liquid biopsy solutions on Ion GeneStudio S5 Systems	
Oncomine Pan-Cancer Cell-Free Assay	A37664
Oncomine Lung Cell-Free Total Nucleic Acid Assay	A35864
Oncomine Lung cfDNA Assay	A31149
Oncomine Breast cfDNA Assay v2	A35865
Oncomine Breast cfDNA Assay	A31183
Oncomine Colon cfDNA Assay	A31182
Tag Sequencing Barcode Set 1-24	A31830
Tag Sequencing Barcode Set 25-48	A31847
Ion GeneStudio S5 Prime System	A38196
Ion GeneStudio S5 Plus System	A38195
Ion GeneStudio S5 System	A38194
Ion 550 Chip Kit	A34538
Ion 540 Chip Kit	A27766
Ion 530 Chip Kit	A27764
Ion Chef System	4484177
Ion 550 Kit-Chef	A34541
Ion 540 Kit-Chef	A30011
Ion 510 & Ion 520 & Ion 530 Kit-Chef	A34461
Liquid biopsy solutions on Ion Torrent Genexus System	
Oncomine Precision Assay GX	A46291
Ion Torrent Genexus System	A45727
Ion Torrent Genexus GX5 and Coupler	A40269
Ion Torrent Genexus GX5 Starter Pack-AS	A40279
Oncomine informatics	
Ion Reporter Server System	4487118
Oncomine Reporter	A33109

Find out more at thermofisher.com/liquidbiopsy-ngs