



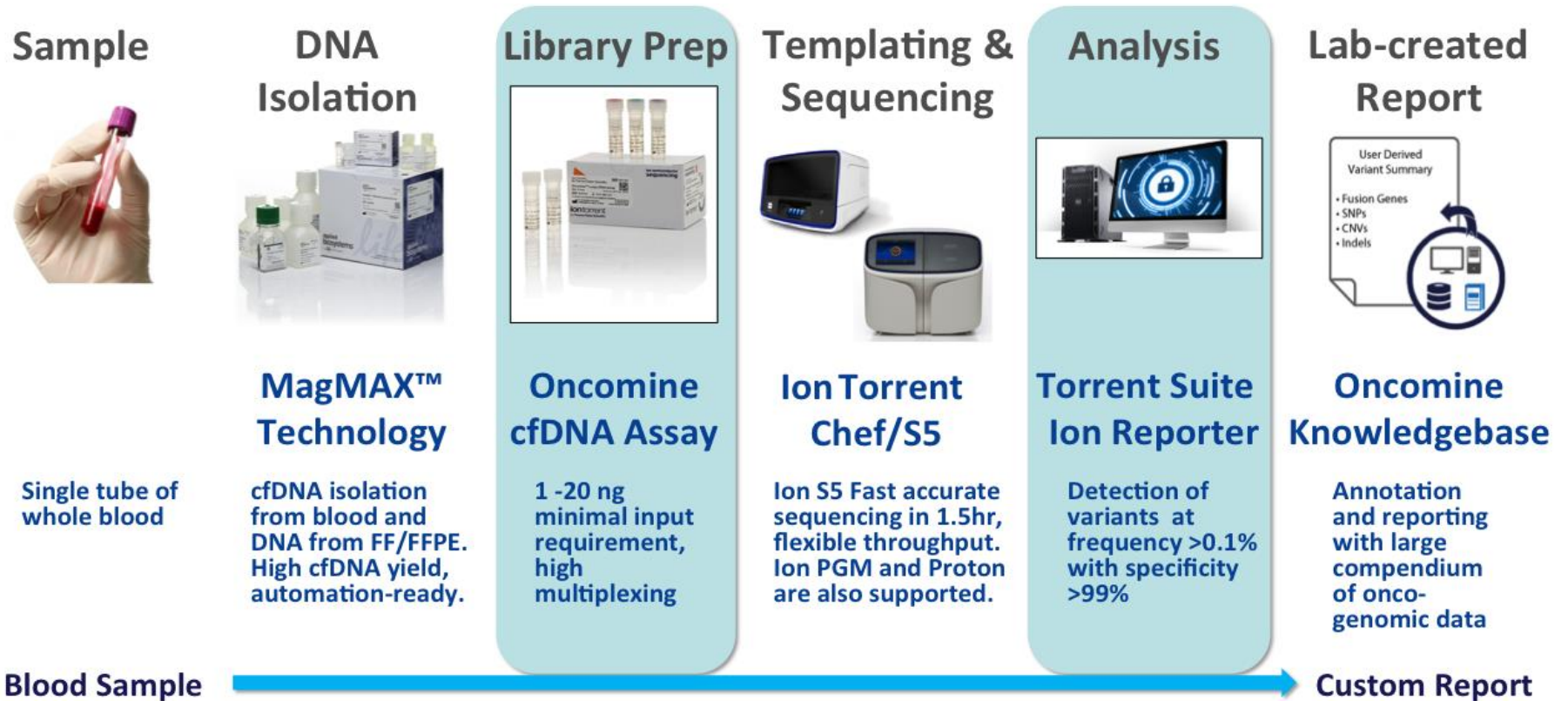
**ThermoFisher**  
SCIENTIFIC

# Advancing Liquid Biopsy with NGS Solutions

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# Workflow for cfDNA Analysis on Ion Torrent™ S5XL NGS Platform



- Core technology for Library Prep consists of an amplification based assay that generates tagged DNA copies. This allows one to identify reads amplified from the same original DNA molecule, and identify molecules containing variants.
- Total process time (from plasma/FFPE specimen to report) as short as 32 hours with a total hand on time of ~4 hours.

# OncoPrint™ cfDNA Assays | Content

- Designed to detect **primary tumor drivers and resistance mutations**
- Reagents, primers, and analysis software to analyze mutations from a **single tube of blood**
- Flexible input amounts and tolerance of sample input variability to achieve **0.1% limit of detection** of SNV hotspots and indels from cfDNA

## OncoPrint™ Lung cfDNA Assay

<i>ALK</i>	<i>MET</i>
<i>BRAF</i>	<i>NRAS</i>
<i>EGFR</i>	<i>PIK3CA</i>
<i>ERBB2</i>	<i>ROS1</i>
<i>KRAS</i>	<i>TP53</i>
<i>MAP2K1</i>	

35 amplicon panel for Lung

Covering key hotspot mutations in 11 genes

169 Hotspot SNVs & indels

## OncoPrint™ Colon cfDNA Assay

<i>AKT1</i>	<i>KRAS</i>
<i>BRAF</i>	<i>MAP2K1</i>
<i>CTNNB1</i>	<i>NRAS</i>
<i>EGFR</i>	<i>PIK3CA</i>
<i>ERBB2</i>	<i>SMAD4</i>
<i>FBXW7</i>	<i>TP53</i>
<i>GNAS</i>	<i>APC</i>

49 amplicon panel targeting multiple cancer types

Covering key hotspot mutations in 14 genes

236 Hotspot SNVs & indels

## OncoPrint™ Breast cfDNA Assay

<i>AKT1</i>	<i>FBXW7</i>
<i>EGFR</i>	<i>KRAS</i>
<i>ERBB2</i>	<i>PIK3CA</i>
<i>ERBB3</i>	<i>SF3B1</i>
<i>ESR1</i>	<i>TP53</i>

26 amplicon panel for Breast

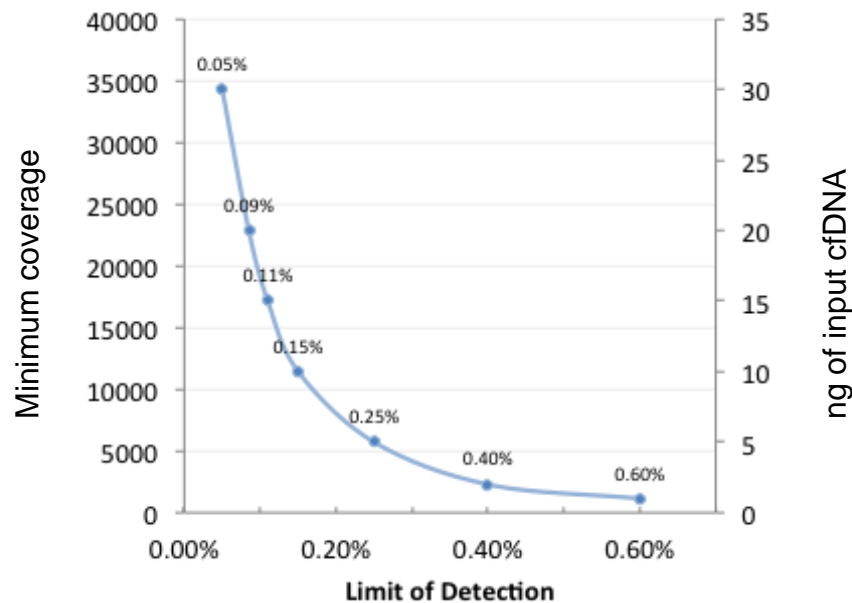
Covering key hotspot mutations in 10 genes

152 Hotspot SNVs & indels

## High Sensitivity and Specificity

	Sample	Sensitivity	Specificity
Lung	0.1% MM	92.2%	99.7%
	0.5% MM	>99.9%	99.6%
Colon	0.1% MM	85.9%	>99.9%
	0.5% MM	>99.9%	>99.9%
Breast	0.1% MM	81.3%	>99.9%
	0.5% MM	>99.9%	>99.9%

## 0.1% Limit of Detection



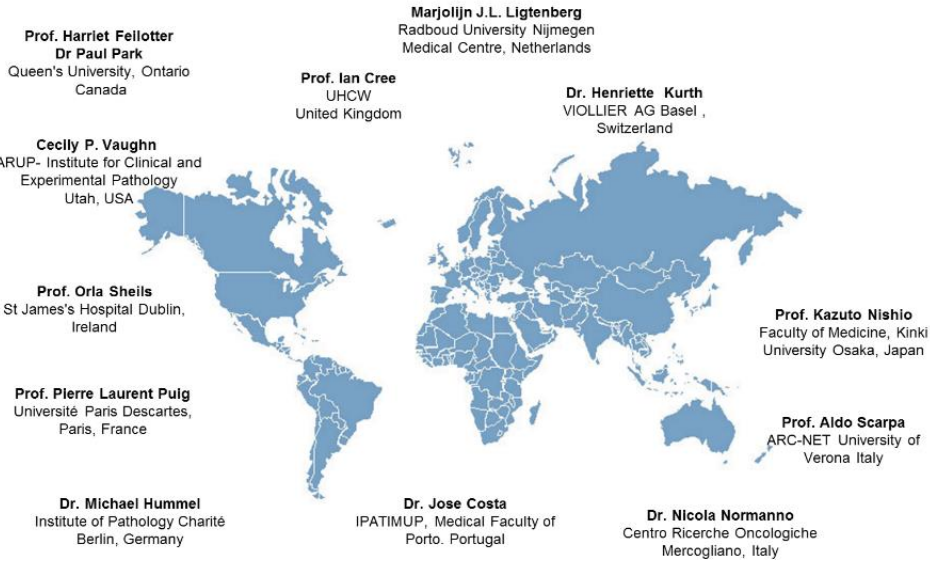
1 ng cfDNA–0.6% LOD  
 5 ng cfDNA–0.25% LOD  
 10 ng cfDNA–0.15% LOD  
**20 ng cfDNA–0.1% LOD**  
 30 ng cfDNA–0.05% LOD

# Horizon BioDiscovery cfDNA Multiplex Reference Set on Lung cfDNA Panel

Sample	<i>EGFR</i> E746_A750d eIELREA	<i>EGFR</i> L858R	<i>EGFR</i> T790M	<i>EGFR</i> V769_D770 insASV	<i>KRAS</i> G12D	<i>NRAS</i> A59T	<i>NRAS</i> Q61K	<i>PIK3CA</i> E545K
0.1% HDX	0.06	0.17	0.06	0.10	0.22	0.17	0.15	0.10
1% HDX	0.72	1.07	0.75	0.74	1.14	1.15	1.15	2.29
5% HDX	4.52	4.86	6.32	3.97	6.34	6.11	6.94	5.29
100% WT	0	0	0	0	0	0	0	0

Numbers in this table indicate the variant frequency called by variant caller analysis plug-in for the 8 somatic variants engineered into the Horizon Multiplex cfDNA Reference Standard Set when analyzed using the OncoPrint Lung cfDNA Assay.

# OncoNetwork consortia – Multi center verification study

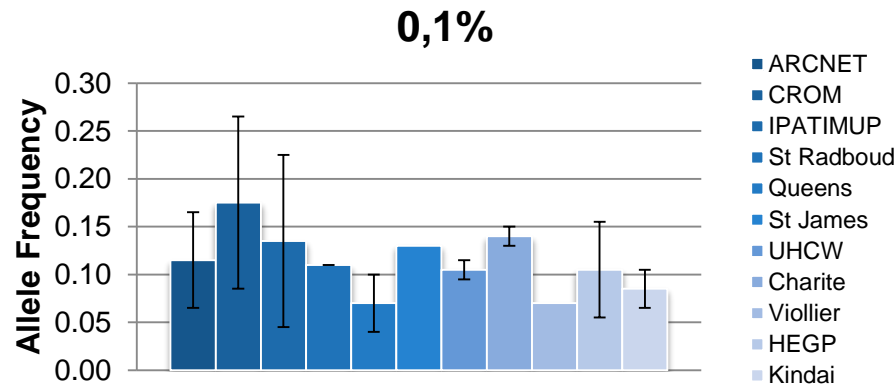
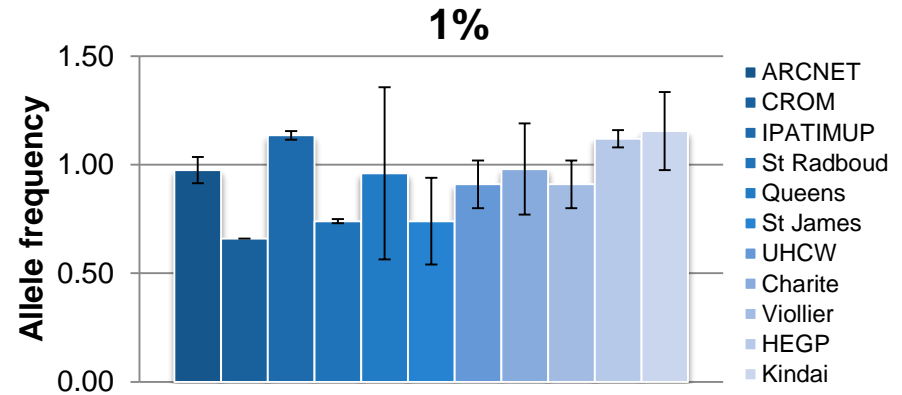
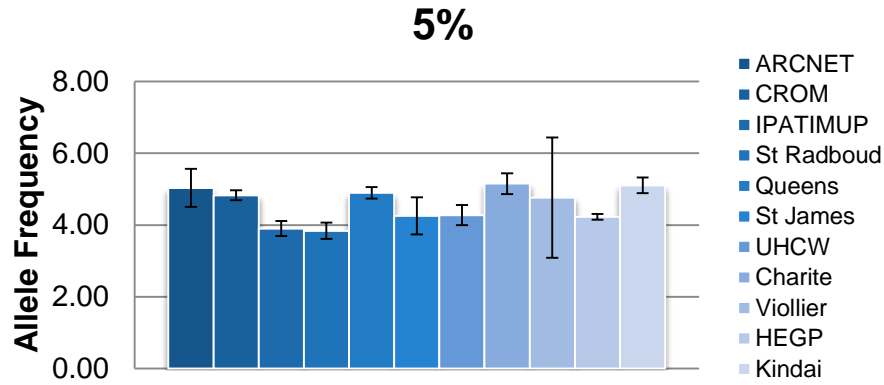


0.1% LOD

Allele frequency	Sensitivity	Specificity
0.1%-5%	94.81%	99.82%
0.1%	83.93%	99.88%

Horizon - Multiplex I cfDNA Reference Standard Set

# OncoNetwork Consortia - EGFR – T790M at 5%, 1% and 0,1% allele frequency



# NEW Oncomine™ Lung and Breast cell free Assays – Available Q3 2017

- Designed to detect **primary tumor drivers and resistance mutations**
- Containing **Fusions and CNVs**

## Enhanced Oncomine Breast cfDNA Assay

<i>AKT1</i>	<i>ESR1</i>	<i>SF3B1</i>
<i>EGFR</i>	<i>FBXW7</i>	<i>TP53</i>
<i>ERBB2</i>	<i>KRAS</i>	
<i>ERBB3</i>	<i>PIK3CA</i>	

Amplicons: **76**

Covering:

- Key hotspot mutations in 10 genes
- **CNVs – CCND1, ERBB2, FGFR1**
- **More complete coverage of TP53 (~80%)**
- **Single library to detect SNVs and CNVs**

SNV LOD down to 0.1% with 20 ng input

Same sensitivity & specificity

## Oncomine™ Lung cell free Total Nucleic Acid Assay

<i>ALK</i>	<i>KRAS</i>	<i>PIK3CA</i>
<i>BRAF</i>	<i>MAP2K1</i>	<i>ROS1</i>
<i>EGFR</i>	<i>MET</i>	<i>TP53</i>
<i>ERBB2</i>	<i>NRAS</i>	

**58 amplicon + 49 Fusion Assays + 3 MET Exon Skipping Assays for new Lung Assay**

Covering:

- Key hotspot mutations in 11 genes
- **Fusions – ALK, RET, ROS1**
- **CNV – MET**
- **MET skipping**

SNV LOD down to 0.1% with 20 ng input

Same Sensitivity & Specificity for SNVs

**Single library from both DNA & RNA**

The content provided herein may relate to products that have not been officially released and are subject to change without notice.



A Mission We're Proud Of

# Mission



**We enable our customers to make the world  
healthier, cleaner and safer**

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# Questions?

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