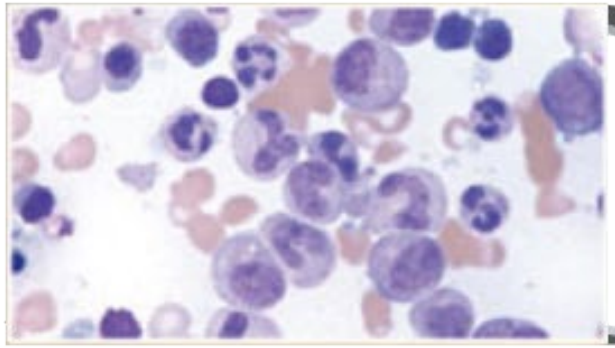
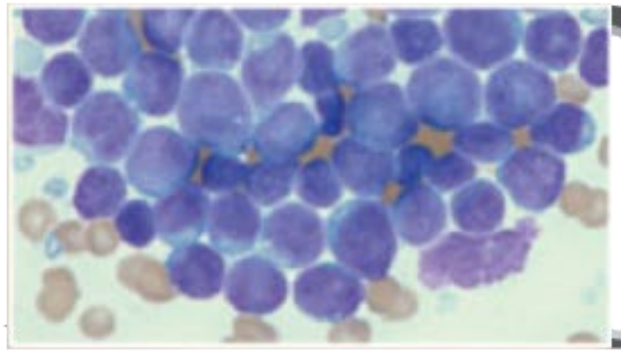
Oncomine Precision Assay on Ion Torrent Genexus System

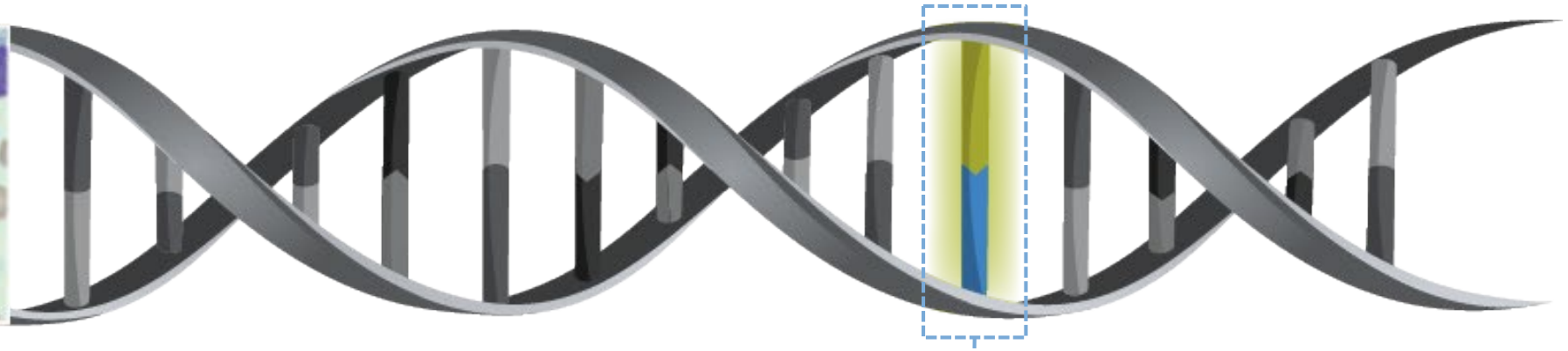
Andy Felton

The Genexus System and its assays are for Research Use Only. Thermo Fisher Scientific is not promoting or encouraging any current diagnostic or clinical use of the Genexus System or any other Research Use Only products discussed in this presentation

Cancer is a Disease of the Genome Caused by its Alterations



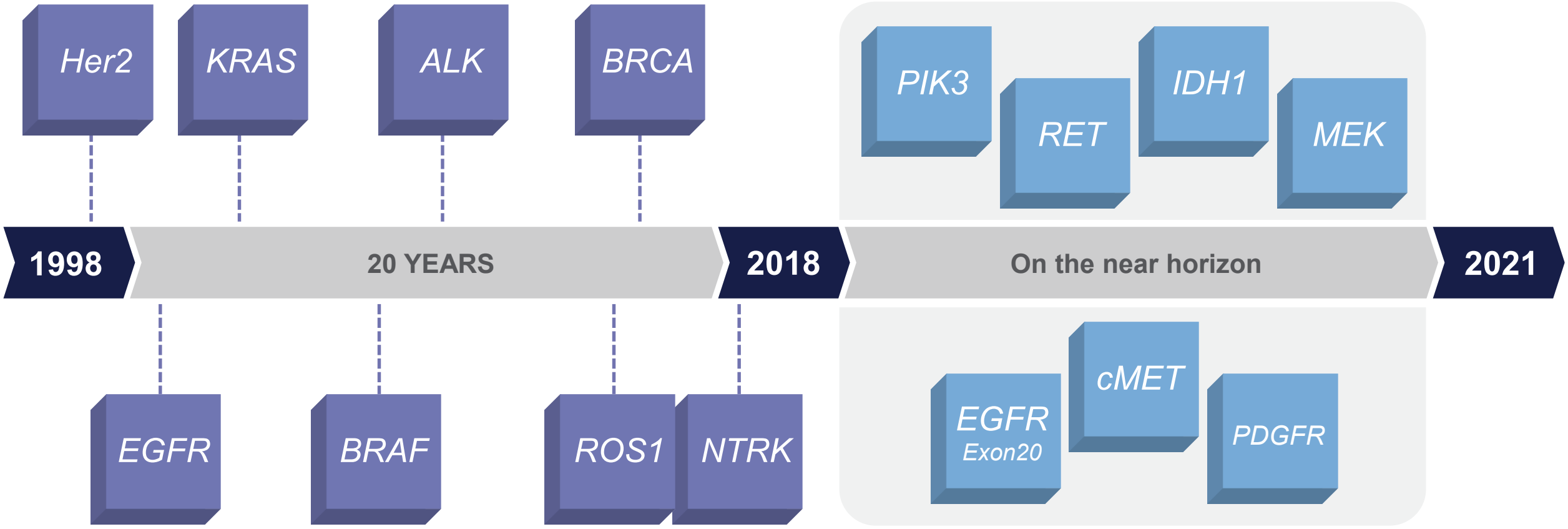


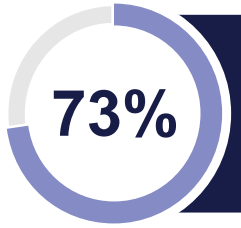


An effective and efficient tool is required to interrogate the alterations that cause cancer

* http://humanbiologylab.pbworks.com/w/page/48192744/Genomes%20%20Finding%20a%20Cure%20for%20Cancer

Biomarker Development is Accelerating





73% of medicines in oncology pipelines have associated biomarkers*

*Personalized Medicine Report 2017 by Personalized Medicine Coalition

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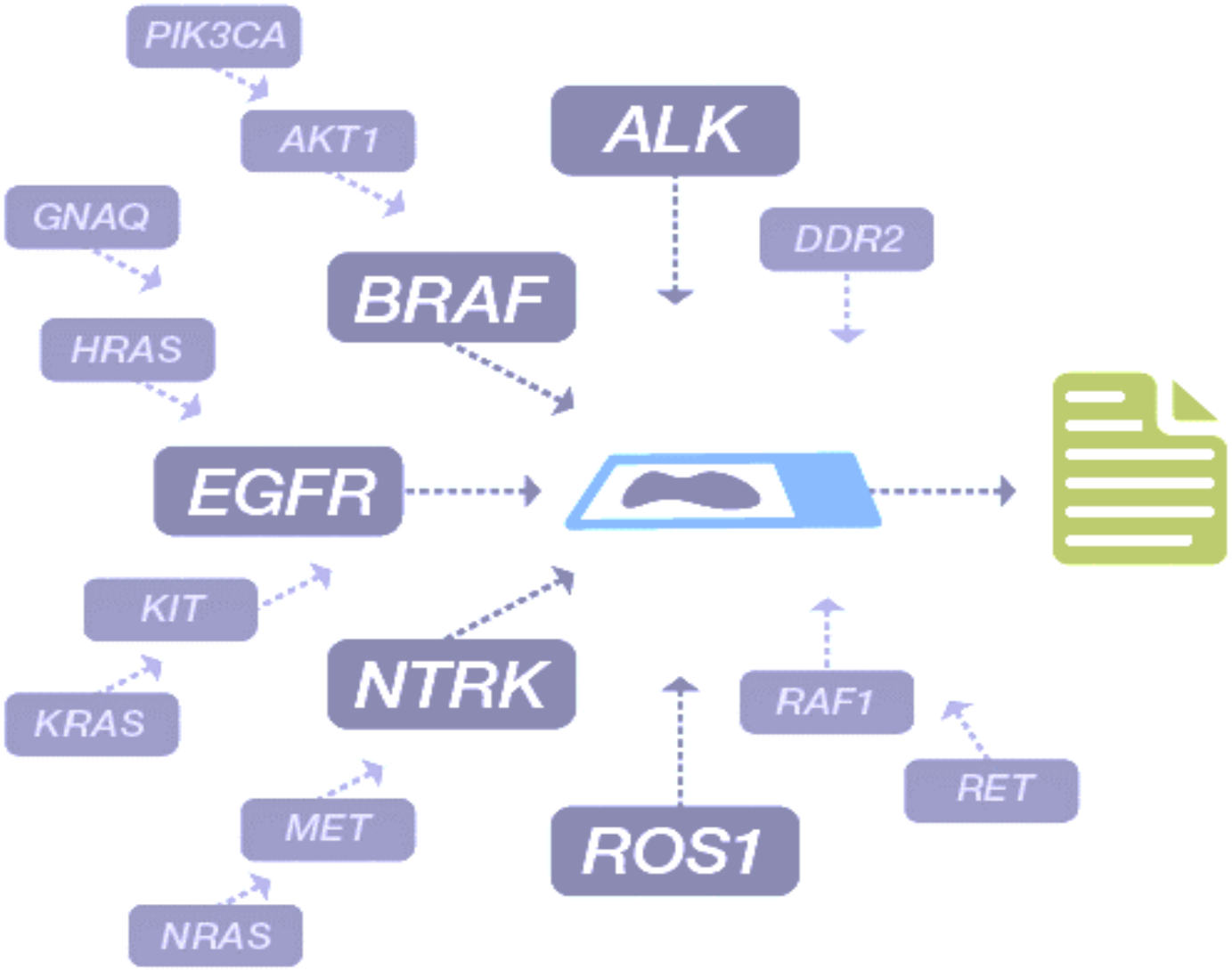
NGS is a Foundation of Precision Oncology Clinical Research

NGS can detect many different types of biomarkers simultaneously from a single sample



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Todays Challenges and Barriers for NGS Implementation in a Broader Lab Spectrum

Too slow

Requires days and often weeks to get the results

Too complex

High level of user expertise required to run NGS

Modular workflows requiring multiple instruments and touchpoints

Too costly

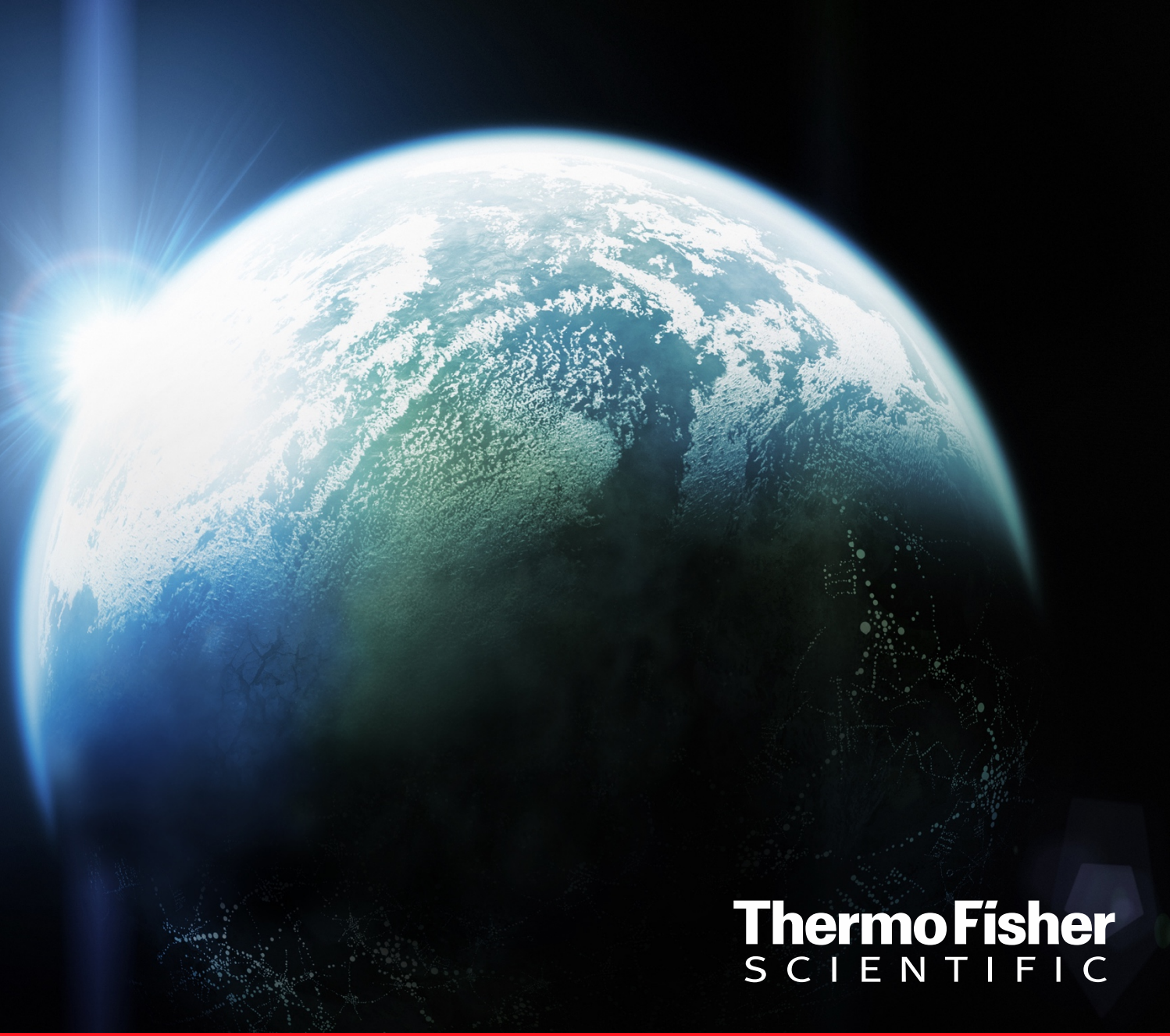
Cost of hiring and training staff

Cost penalty for running small sample batches

Too limited

Tissue requirements / QNS (quantity not sufficient) related failures

A New Day for Genomic Profiling



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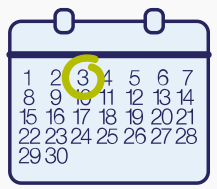
The world leader in serving science

Oncomine Precision Assay on Ion Torrent Genexus System

A new generation solution for genomic profiling

Fast



Single day sample-to-report.

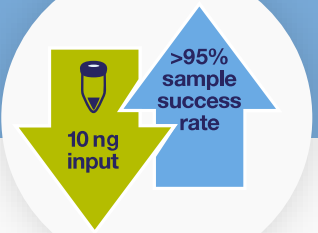
Hands free

2 touch points and only 10 min of hands-on time.

Cost saving

No more need to batch samples across multiple sequencing runs.

Tissue saving



Minimum sample requirement

Maximum results obtained.

*Specimen-to-report workflow will be available after the Ion Torrent™ Genexus™ Purification System and integrated reporting capabilities are added in 2020.
The content provided herein may relate to products that have not been fully validated by Thermo Fisher Scientific and is subject to change without notice.

Introducing Ion Torrent™ Genexus™ System

The world's first turnkey, automated NGS system

Specimen to report in a single day with only two user touchpoints

- **Single-day turnaround time** allowing you to provide IHC and NGS results at the same time
- **Automated** sample purification, library prep, sequencing, and analysis reporting helps increase reproducibility and efficiency, while reducing personnel costs
- **Flexibility of economically running few or one sample** reduces the need for batching and helps you to deliver results faster then ever before
- System and consumables manufactured at a **facilities registered with FDA** and **ISO 13485 certified**
- **Research applications include** oncology & inherited, with reproductive health and infectious to come in the future

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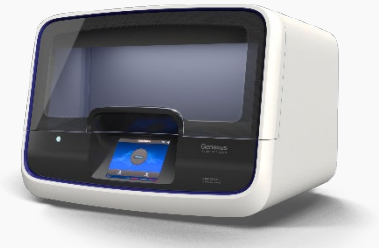
Genexus System—Tomorrow's Specimen-to-Report NGS Workflow

- FFPE tissue
- Frozen tissue
- Bone marrow
- Whole blood
- PBL
- Urine
- Saliva

Genexus Software

Nucleic acid purification and quantitation*

Ion Torrent™ Genexus™
Purification System (Available 2020)



2 hour turnaround time

12 FFPE (DNA and RNA)

6 Plasma

Library preparation to variant interpretation

Report*

Ion Torrent™ Genexus™
Integrated Sequencer (Available November 2019)

Ion Torrent™ GX5™ Chip:
12–15M reads/lane

14 hours for a single-lane run

(approx. 24 to 30 hours for full chip)

Up to 32 Samples per run

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Genexus Software—End-To-End Integration from Specimen to Report

Integrated

Fully integrated solution enabling specimen-to-report workflow; no Ion Reporter server required

Easy to use

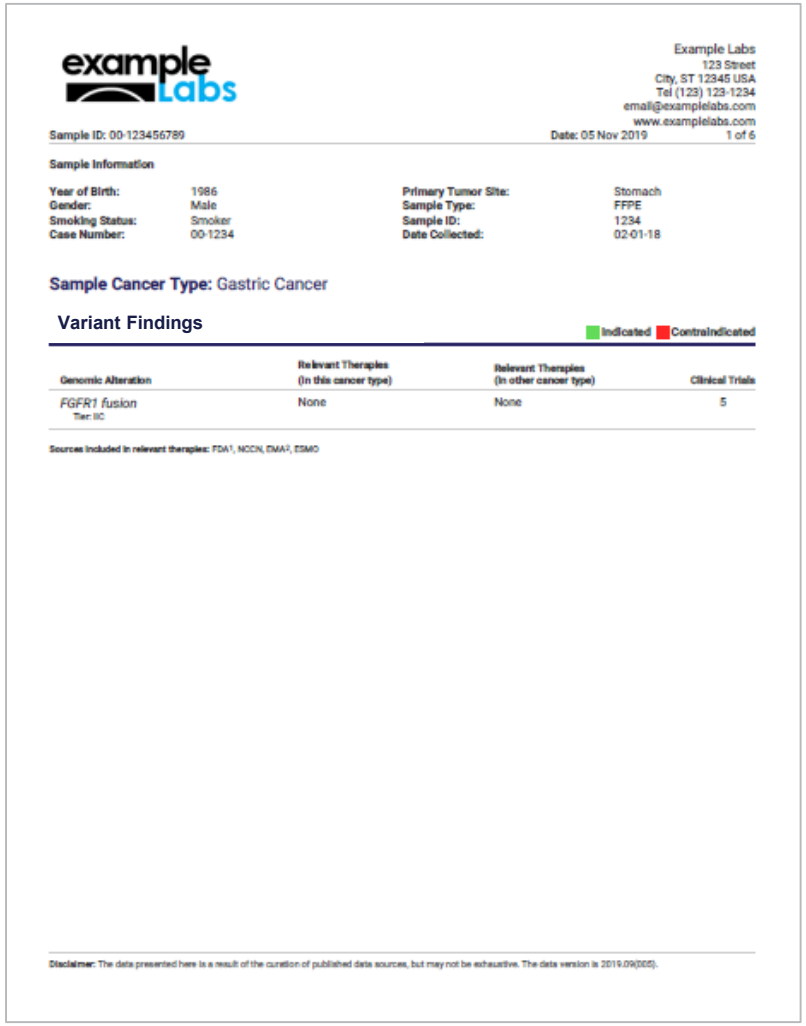
Simplified, new user experience helps minimize the learning curve and human error

Robust

Benchmarks on variant calling accuracy

Flexible

Option to choose between integrated analysis on instrument or analysis on IR server or cloud



example Labs

Example Labs
123 Street
City, ST 12345 USA
Tel (123) 123-1234
email@examplelabs.com
www.examplelabs.com

Sample ID: 00-123456789 Date: 05 Nov 2019 1 of 6

Sample Information

Year of Birth:	1986	Primary Tumor Site:	Stomach
Gender:	Male	Sample Type:	FFPE
Smoking Status:	Smoker	Sample ID:	1234
Case Number:	00-1234	Date Collected:	02-01-18

Sample Cancer Type: Gastric Cancer

Variant Findings — Indicated Contraindicated

Genomic Alteration	Relevant Therapies (in this cancer type)	Relevant Therapies (in other cancer type)	Clinical Trials
FGFR1 fusion Tier: IIC	None	None	5

Sources included in relevant therapies: FDA[1], NCCN, EMA[2], ESMO

Disclaimer: The data presented here is a result of the curation of published data sources, but may not be exhaustive. The data version is 2019.09(005).

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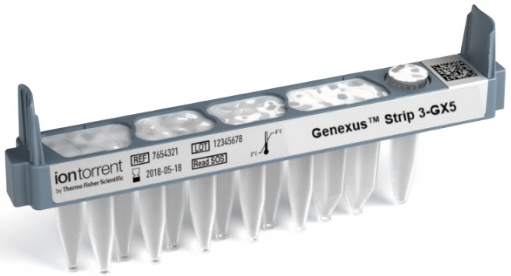
Genexus™ Integrated Sequencer Maximizes Flexibility

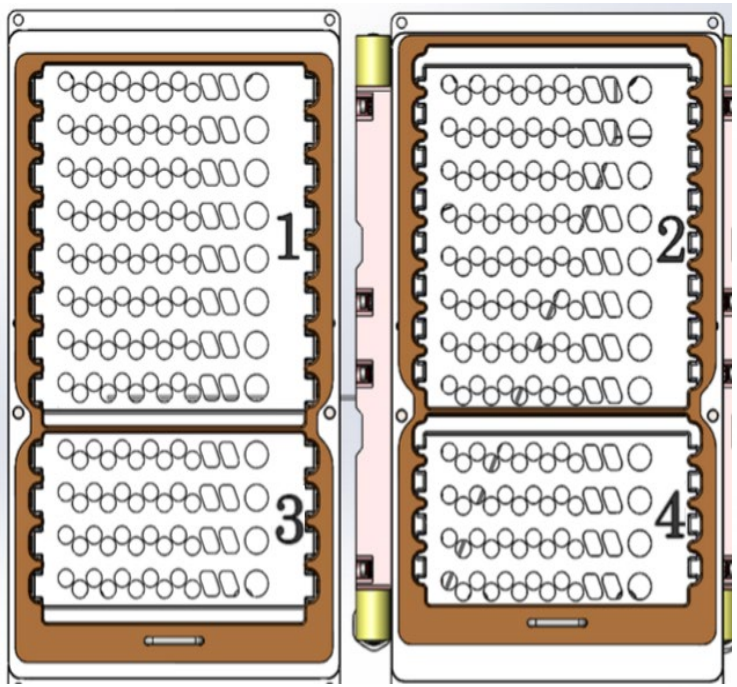
- **Up to four different assays** prepared and sequenced simultaneously in a single run
- **Multiplexing capability** of up to 32 library preparations in a single run

  32 single-pool | 16 two-pool | 8 four-pool
- **Two week** on-instrument chip and reagent stability
- **Assay and throughput flexibility** facilitated by strips and multi-lane sequencing chip
- **Minimized consumable footprint** reduces required storage space

Library strips
1 and 2

Templating strips
3 and 4





Strips 1 through 4, with descriptors for library chemistry or chip type, are installed in corresponding reagent bays on the deck of the Integrated Sequencer

Genexus™ Conical Bottles: Multi-use

Ion GX5 Chip:
12–15M reads/lane

Genexus™ Cartridge

Workflow Micro-costing Study

Comparison of 4 Workflows

- DL8/Chef/S5
- Manual Library Prep/Chef/S5
- Manual Library Prep/ MiSeq
- Genexus

Users

- 2 novice users and 3 expert AmpliSeq users

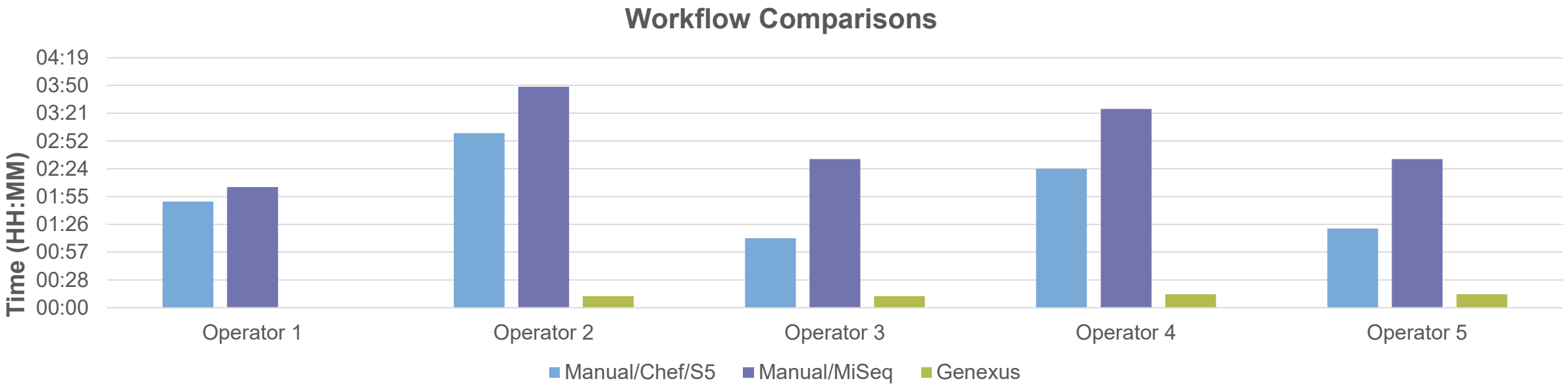
Training

- All users received overview training on ILMN workflow
- All users are novice users of Genexus

Technical Decisions

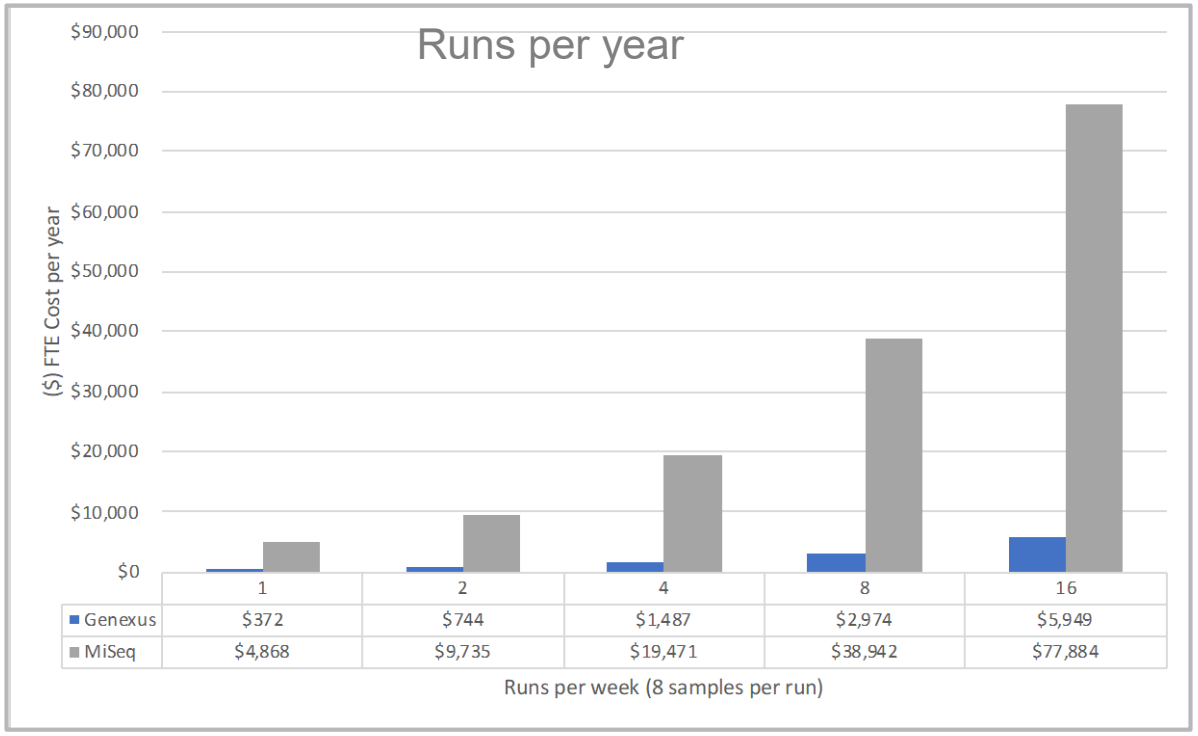
- 8 libraries were created for each workflow by each operator
- Decision not to use Biomek – customers will most likely not use Biomek for only 8 libraries

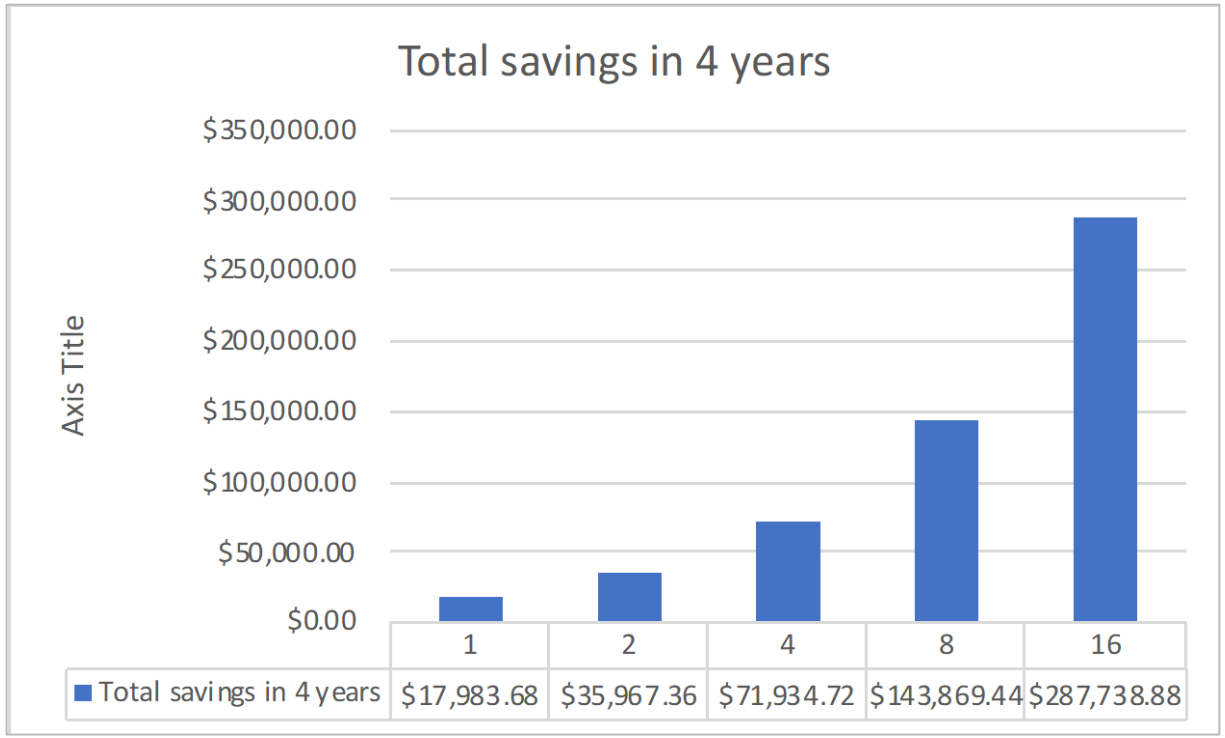
Comparisons – Total Hands-On Time (Does not include Sample QC)



Users	Manual/Chef/S5	Manual/MiSeq	Genexus
Operator 1	01:50	02:05	N/A
Operator 2	03:01	03:49	00:12
Operator 1	01:12	02:34	00:12
Operator 2	02:24	03:26	00:14
Operator 3	01:22	02:34	00:14
Average	**01:56**	**02:54**	**00:13**

Workflow Micro-costing Study Results





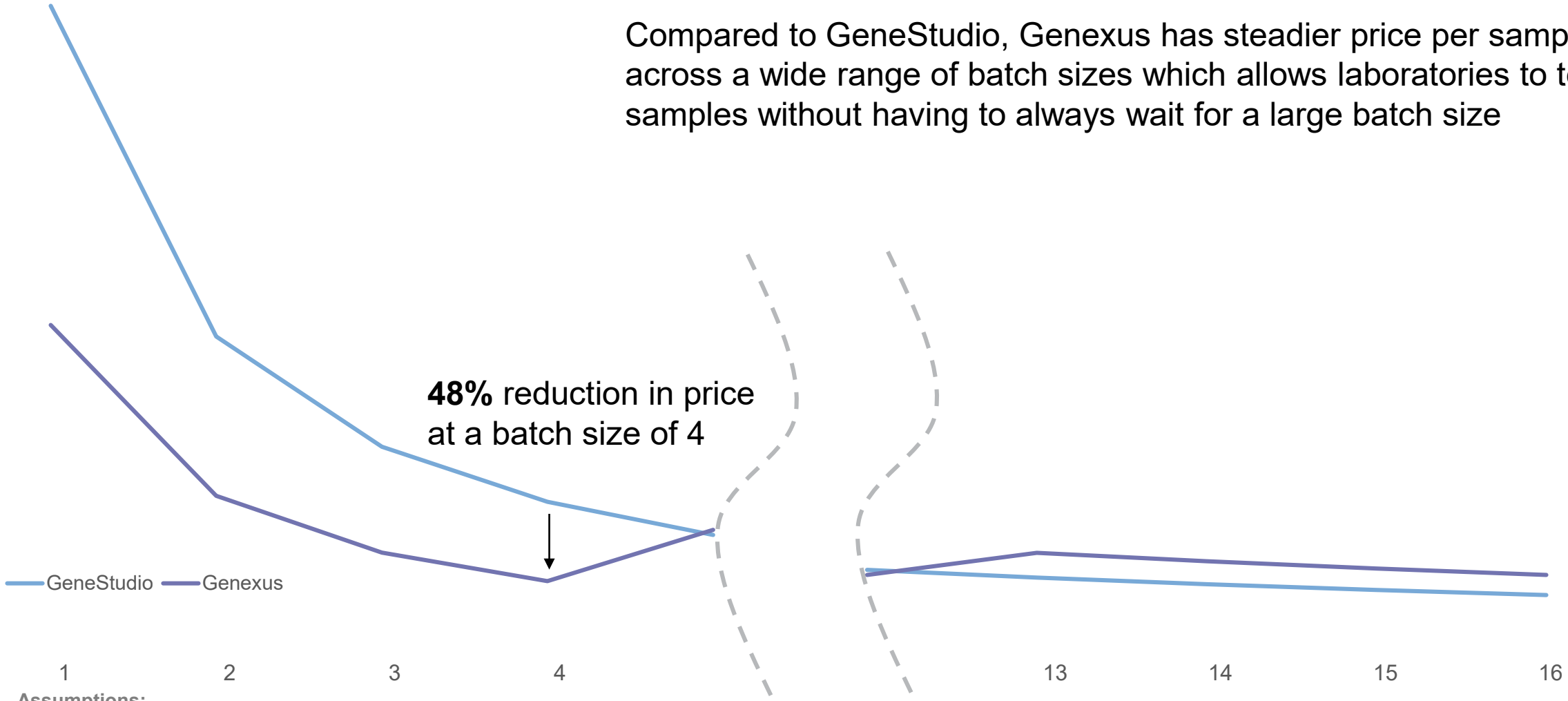
- Samples: 8 samples per run (OFA)
- FTE rate: $33/hr (Glassdoor and Indeed rates for Clinical Lab Tech)
- Genexus FTE cost per run: $7.15
- MiSeq FTE cost per run: $93.61 (short of data analysis FTE costs)

Difference between Genexus and MiSeq FTE cost

- 52 weeks
- 4 year based on amortization of capital assets

Genexus Is Cost Effective at Low Batch Sizes

Compared to GeneStudio, Genexus has steadier price per sample across a wide range of batch sizes which allows laboratories to test samples without having to always wait for a large batch size



Assumptions:

- For Genexus, per sample pricing calculated using list prices for Oncomine Precision Assay with associated Genexus consumables (running FFPE samples)
- For GeneStudio, per sample pricing calculated using list prices for Oncomine Focus Assay with associated GeneStudio S5 consumables (Ion Chef automated library preparation)

Genexus Integrated Sequencer and Assay Turnaround Times

Metric	Assay	1 Lane	2 Lanes	3 Lanes	4 Lanes
Output in Reads (M)		12-15	24-30	36-45	48-60
Turnaround Time (*Batch Size*) Nucleic Acid to Oncomine Report	Custom AmpliSeq 1 Pool Assay	14 (*4*)	NA	NA	24 (*16*)
	Oncomine Comprehensive Assay v3 (DNA & RNA workflow)	18.5 (*1*)	20.5 (*3*)	27 (*4*)	29.5 (*6*)
	Oncomine Precision Assay (cfTNA workflow)	16.5 (*1*)	17.5 (*2*)	20 (*3*)	22.5 (*4*)
	Oncomine Precision Assay (FFPE workflow)	16.5 (*3*)	21 (*6*)	26 (*9*)	28.5 (*12*)
	Oncomine TCR Beta-LR Assay	20 (*4*)	24 (*8*)	28.5 (*12*)	31 (*16*)

Oncomine Precision Assay on Ion Torrent Genexus System

Maximizes your ability to detect relevant variants

Curated pan-cancer content

- Mutations, CNVs, and fusion variant types across 50 key genes
- Tumor suppressors, drivers, and resistance variants

Tissue and plasma samples

- One test, one workflow, multiple sample types
- Maximizes the number of tumors that can be profiled

Molecular tagging

- Enhanced low-level variant detection
- Key for liquid biopsy testing

FusionSync™ Detection Technology

- Sensitive and specific—targeted isoform designs
- Novel fusion detection

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

The Oncomine Precision Assay Content

The Oncomine Precision Assay content is carefully curated to include all relevant targets and targets of emerging importance in precision oncology clinical research..

- 50 genes and 2,769 unique variants
- Mutations (45), CNVs (14), and fusion variants (19),
- Pan cancer span with NSCLC focus
- 218 potential resistance mutations across 22 genes

Oncomine Precision Assay Gene Content

DNA hotspots

AKT1	ESR1	MAP2K2
AKT2	FGFR1	MET
AKT3	FGFR2	MTOR
ALK	FGFR3	NRAS
AR	FGFR4	NTRK1
ARAF	FLT3	NTRK2
BRAF	GNA11	NTRK3
CDK4	GNAQ	PDGFRA
CDKN2A	GNAS	PIK3CA
CHEK2	HRAS	PTEN
CTNNB1	IDH1	RAF1
EGFR	IDH2	RET
ERBB2	KIT	ROS1
ERBB3	KRAS	SMO
ERBB4	MAP2K1	TP53

CNV

ALK
AR
CD274
CDKN2A
EGFR
ERBB2
ERBB3
FGFR1
FGFR2
FGFR3
KRAS
MET
PIK3CA
PTEN

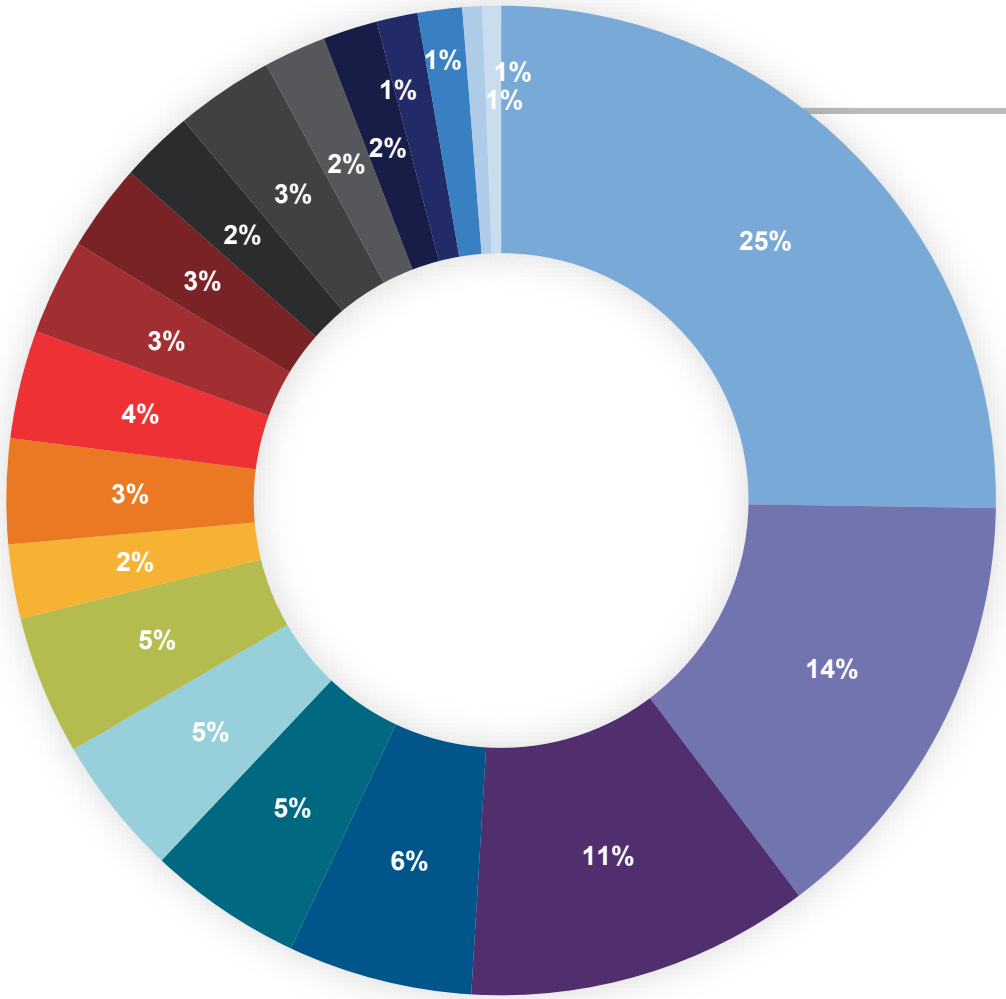
Inter-genetic fusions

ALK	**NTRK2**
BRAF	**NTRK3**
ESR1	NUTM1
FGFR1	**RET**
FGFR2	ROS1
FGFR3	RSPO2
MET	RSPO3
NRG1	
NTRK1	

Intra-genetic fusions

AR
BRAF
EGFR
MET

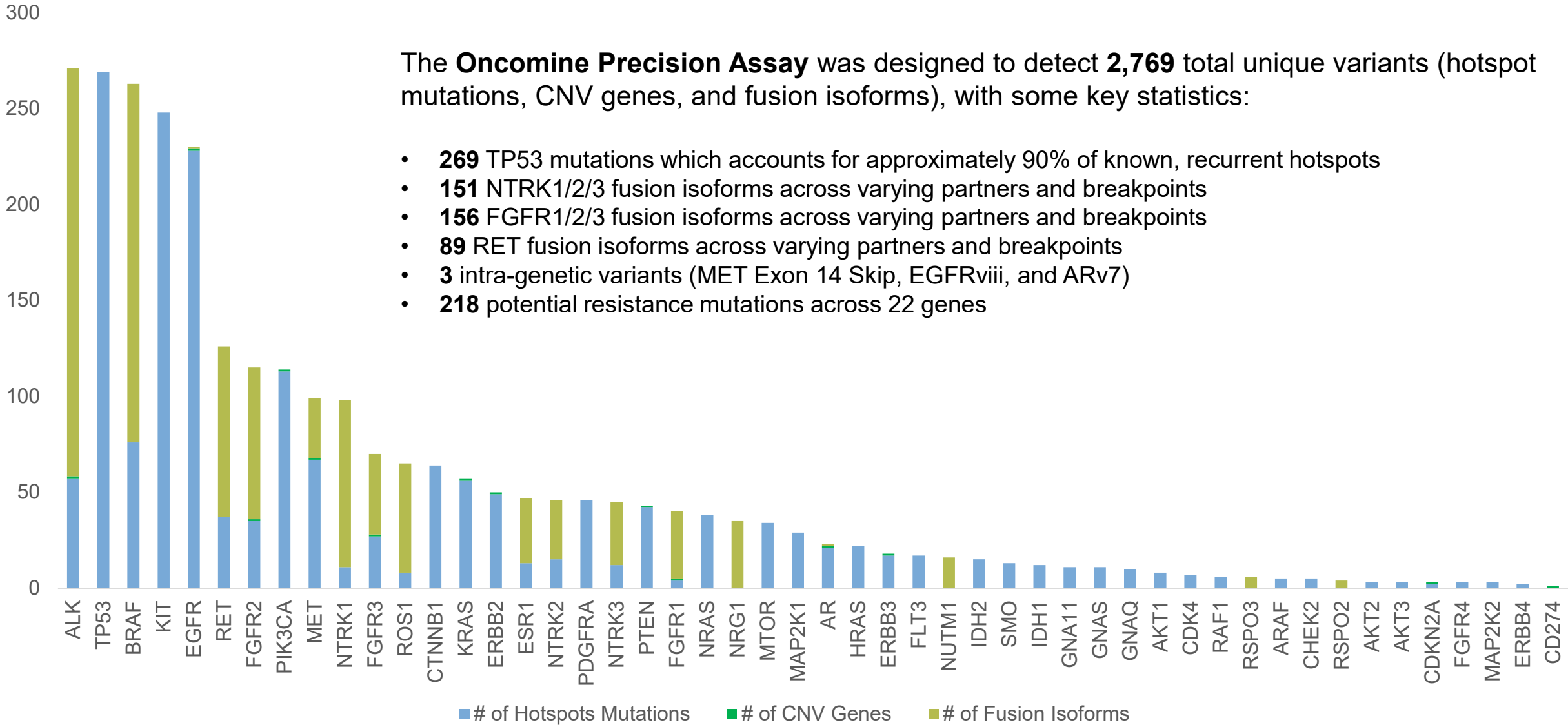
Pan-Cancer Clinical Research Application of OPA



- Non-Small Cell Lung Cancer
- Breast Cancer
- Melanoma
- Gastric Cancer
- Bladder Cancer
- Head and Neck Cancer
- Pancreatic Cancer
- Thyroid Cancer
- Soft Tissue Sarcoma
- Small Cell Lung Cancer
- Unspecified Solid Tumor
- Colorectal Cancer
- Kidney Cancer
- Ovarian Cancer
- Esophageal Cancer
- Endometrial Cancer
- Liver Cancer
- Glioblastoma
- Gastrointestinal Stromal Tumor
- Cervical Cancer

As expected, NSCLC continues to be the most “biomarker-rich” indication

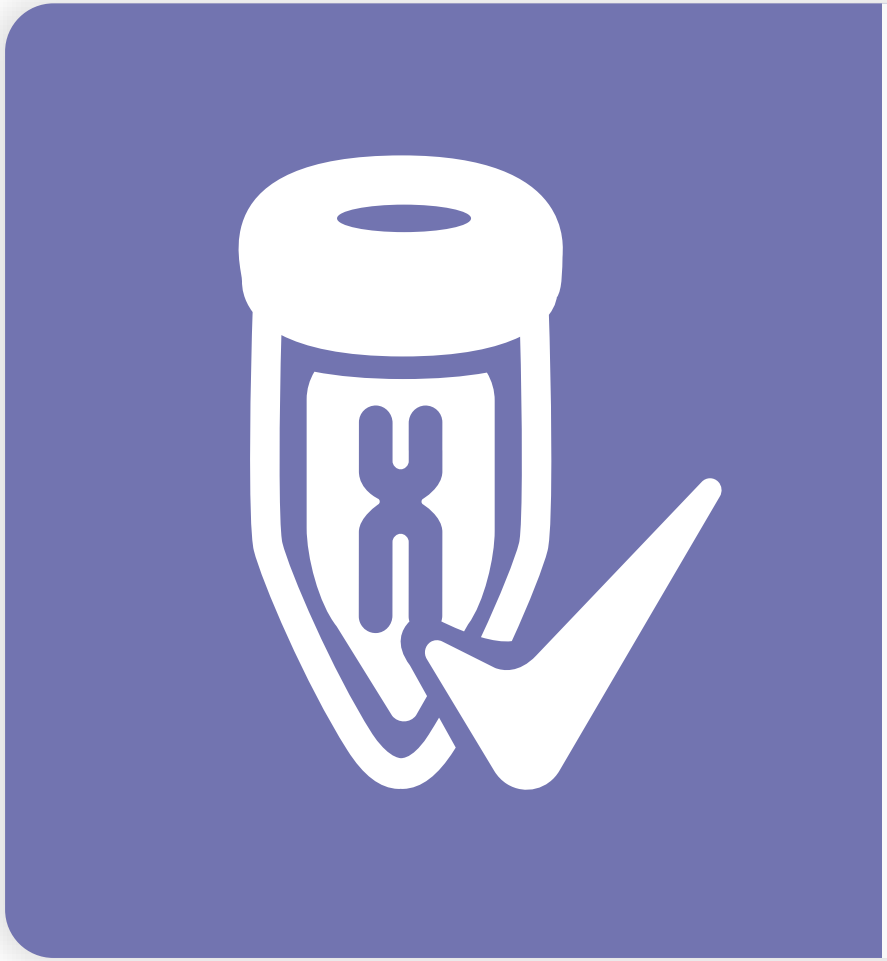
Oncomine Precision Assay Design Details



The **Oncomine Precision Assay** was designed to detect **2,769** total unique variants (hotspot mutations, CNV genes, and fusion isoforms), with some key statistics:

- **269** TP53 mutations which accounts for approximately 90% of known, recurrent hotspots
- **151** NTRK1/2/3 fusion isoforms across varying partners and breakpoints
- **156** FGFR1/2/3 fusion isoforms across varying partners and breakpoints
- **89** RET fusion isoforms across varying partners and breakpoints
- **3** intra-genetic variants (MET Exon 14 Skip, EGFRviii, and ARv7)
- **218** potential resistance mutations across 22 genes

Benefits of Oncomine Fusion Detection Technology



Generally, there are **two** key features for optimal fusion detection:

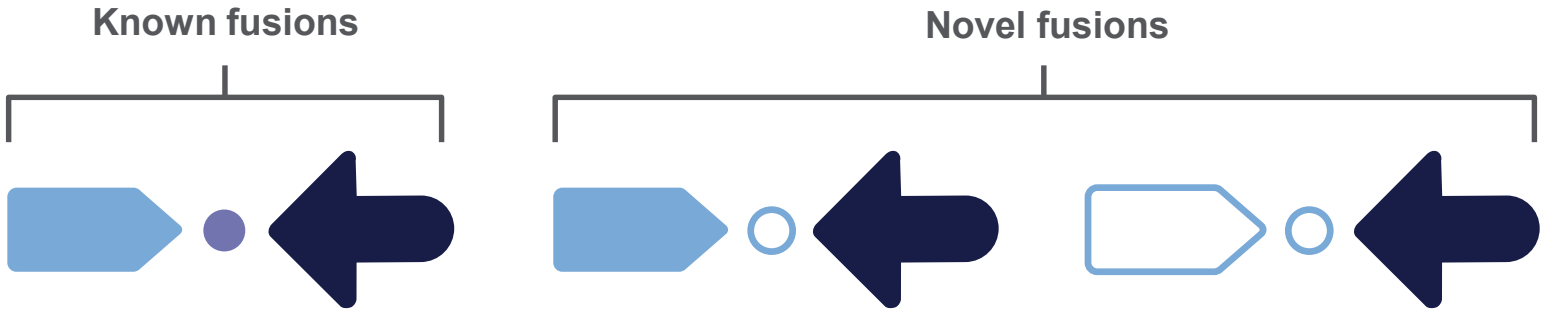
1. Performance of fusion detection with low input samples / low level transcripts
2. Ability to detect novel fusions for driver genes (e.g. *NTRK* and *FGFR*)

Many similar technologies emphasize #2 above but ignore #1.

With FusionSync™ detection, we address **BOTH #1 and #2**

FusionSync Detection Technology

FusionSync Detection Technology is a synchronous approach that combines three methods for sensitive, specific, and broad detection of known and novel fusions



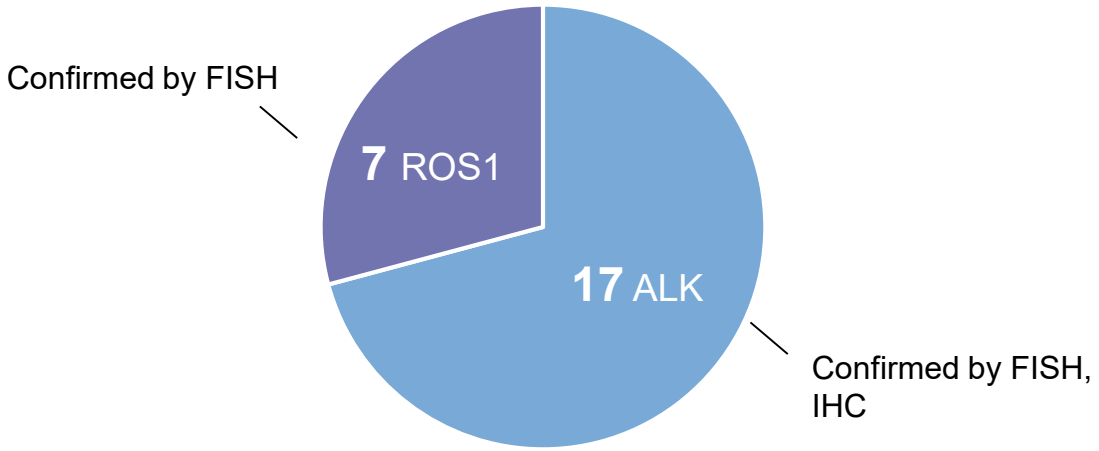
Method	*Targeted*	*Non-fargeted*	*Exon* filing imbalance*
Driver	✓ Known	✓ Known	✓ Known
Partner	✓ Known	✓ Known	○ Unknown
Breakpoint	✓ Known	○ Unknown	○ Unknown

** Available for ALK, FGFR1, FGFR2, FGFR3, NTRK1, NTRK2, NTRK3, and RET fusion drivers*

FusionSync™ Detection Using Genexus and Oncomine Precision Assay

Detection of ALK and ROS1 fusions

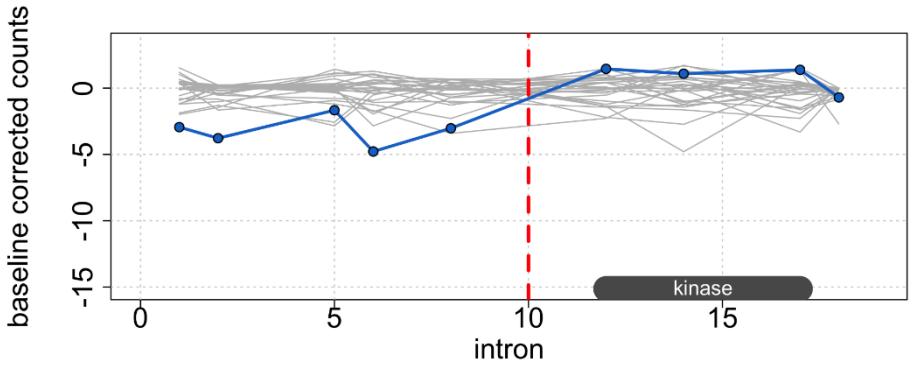
24 NSCLC FFPE samples with orthogonally confirmed ALK or ROS1 fusions were tested:



100% agreement in detecting ALK and ROS1 fusions using Oncomine Precision Assay and Genexus

Testing was performed at multiple internal R&D laboratories as part of product verification testing. Samples were run at different plexy levels. Additional verification and validation testing will be performed using the Oncomine Precision Assay and FFPE samples; therefore final performance values can change.

Detection of RET fusion using **exon tiling imbalance**



Fusion Driver Detected	Predicted Breakpoint Range	Imbalance Score *(threshold)*	P-Value *(threshold)*
RET	ex8-ex12	2.12 *(> 1.65)*	3e-03 *(< 0.05)*

Testing on LC/2ad cell line that includes CCDC6-RET fusion. Blue line indicates normalized read counts of tiled amplicons across RET, showing a differential expression from the 3' (right) to 5' (left) end of the gene. Collection of grey lines indicate RET expression from fusion wild-type samples, to be used as a baseline comparison to sample RET expression measurement (blue line). Red dotted line indicates predicted break point of RET fusion

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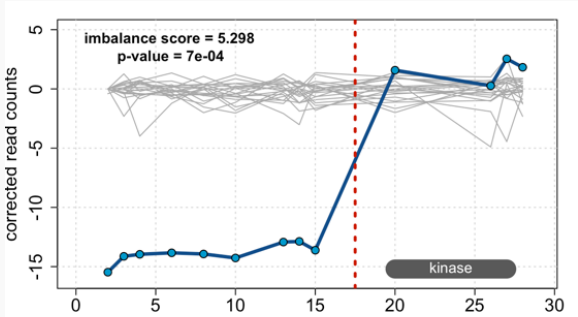
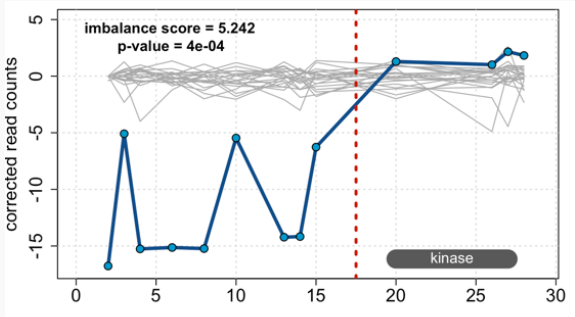
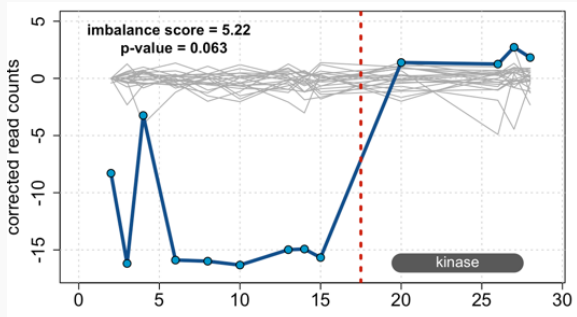
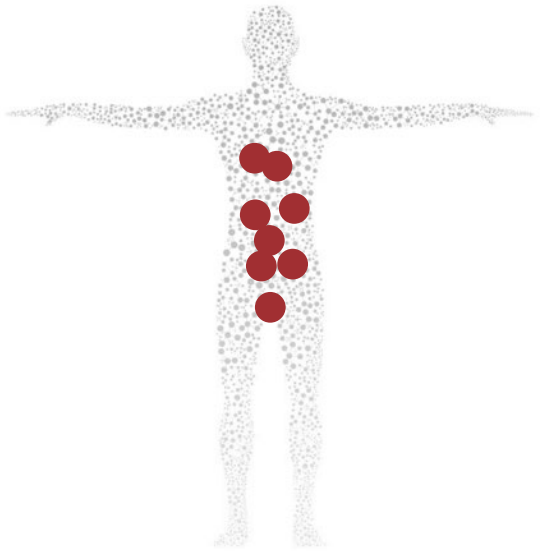
ALK Example: FusionSync™ Detection Using Targeted and Novel Detection Methods

	HIP1-ALK.H21A20	***KIF5B-ALK***.K17A20	***EML4-ALK***.E20A20
Targeted isoform detection	**Isoform read count:** 373 **Isoform molecular count:** 3	**Isoform read count:** 602 **Isoform molecular count:** 10	**Isoform read count:** 671 **Isoform molecular count:** 12
Novel detection using imbalance			

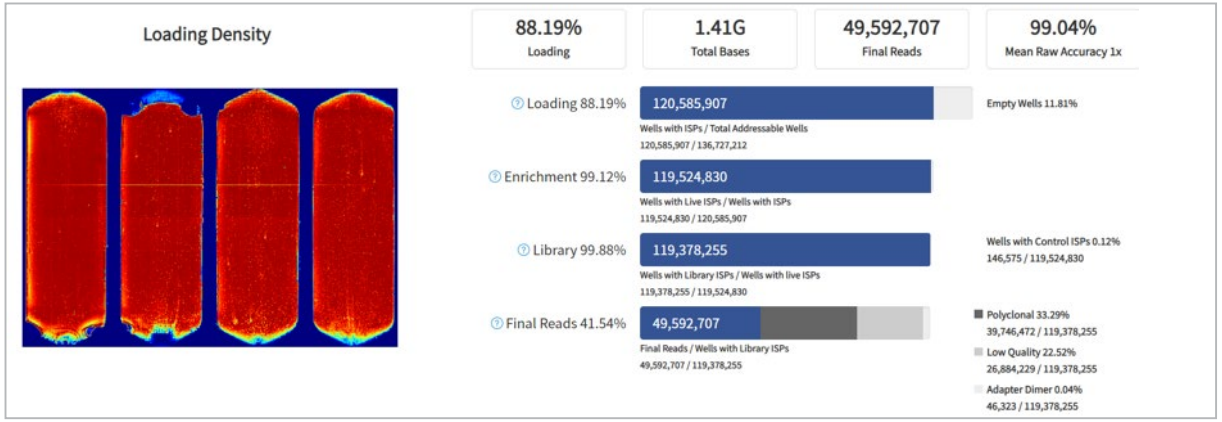
FFPE Samples Successfully Tested Using Oncomine Precision Assay

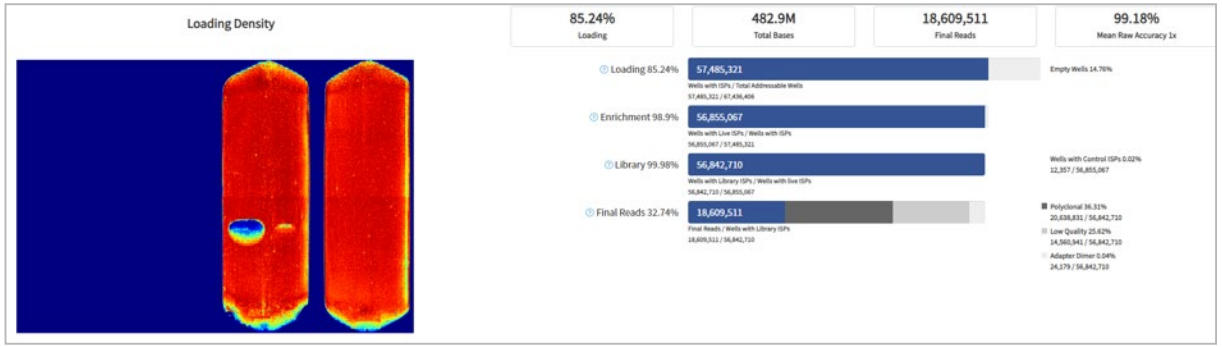
Uncharacterized FFPE samples were included in internal verification testing using the **Oncomine Precision Assay** and **Genexus Integrated Sequencer**. Results demonstrated successful sequencing of different tumor and biopsy types.



Cancer Type
Bladder
Breast (x2)
Kidney (x2)
Ovarian
Stomach (x2)
Testis
Uterus (x2)

	Cancer Type
FNA	Lung
	Lung
	Lung
CNB	Breast
	Breast
	Lung





**NOTE: Samples were not orthogonally tested, therefore variant results are not presented*

NOTE: *Testing was performed at multiple internal R&D laboratories as part of product verification testing. Samples were run at different plexy levels. Additional verification and validation testing will be performed using the Oncomine Precision Assay and FFPE samples; therefore final performance values can change.*

Performance of Oncomine Precision Assay on Liquid Biopsy Controls

Following are results from internal verification testing using the **Oncomine Precision Assay** and **Genexus Integrated Sequencer** with liquid biopsy controls.

Sample	Description	Range	Variant Type	# of Variants Per Sample	# of Sample Replicates	Sensitivity	PPV
Internal Liquid Biopsy Control	Synthetic control across 24 genes	0.33% ± 0.17% SD AF*	SNV	67	32	89.2%	100%
			INDEL	4	32	100%	100%
Internal Liquid Biopsy Control	Synthetic control that contains *EGFR* amplification	1.16X fold change	CNV	1	32	100%	100%
Internal Liquid Biopsy Control	Synthetic control that contains *MET* exon 14 skip	1% Tri-Fusion and *MET* Exon 14 Skip in Total RNA	FUSION	1	32	97.7%	96.4%

NOTE: *Testing was performed at multiple internal R&D laboratories as part of product verification testing. Samples were run at different plexy levels. Additional verification and validation testing will be performed using the Oncomine Precision Assay and FFPE controls; therefore final performance values can change.*
** indicates observed allele frequency using Oncomine Precision Assay on Genexus.*

Key Genes and Variant Types Detected with Cell Lines and FFPE Samples

The following are key variants that were detected using the Oncomine Precision Assay and from cell lines and FFPE samples:

Gene	Variant	Variant Type	Sample Type
EGFR	p.A767_V769dup	Insertion	FFPE
EGFR	p.E746_S752delinsV	Deletion	FFPE
KRAS	G12D	SNV	FFPE
ERBB2 (HER2)	Amplification	CNV	FFPE
ROS1	Fusion	FUSION	FFPE
PTEN	Loss	CNV	Cell Line

NOTE: *Testing was performed at multiple internal R&D laboratories as part of product verification testing. Samples were run at different plexy levels. Additional verification and validation testing will be performed using the Oncomine Precision Assay and FFPE samples; therefore final performance values can change.*

Detection of *ALK* and *ROS1* Fusions in FFPE Samples

Sample #	FISH	IHC	OPA on Genexus	Agreement	Imbalance	P-Value	Breakpoint	Concordance
1	*ALK*	*ALK*	*EML4-ALK*	YES	5.313	0.0007	exon15-exon20	YES
2	*ALK*	*ALK*	*EML4-ALK*	YES	3.969	0.0007	exon14-exon15	YES
3	*ALK*	*ALK*	*EML4-ALK*	YES	5.021	0.0007	exon15-exon20	YES
4	*ALK*	*ALK*	*EML4-ALK*	YES	5.313	0.0007	exon15-exon20	YES
5	*ALK*	*ALK*	*EML4-ALK*	YES	5.273	0.0007	exon15-exon20	YES
6	*ALK*	*ALK*	*EML4-ALK*	YES	3.033	0.0007	exon10-exon13	YES
7	*ALK*	*ALK*	*EML4-ALK*	YES	5.189	0.0007	exon15-exon20	YES
8	*ALK*	*ALK*	*EML4-ALK*	YES	5.276	0.0007	exon15-exon20	YES
9	*ALK*	*ALK*	*EML4-ALK*	YES	5.034	0.005	exon15-exon20	YES
10	*ALK*	*ALK*	*EML4-ALK*	YES	5.126	0.0007	exon15-exon20	YES
11	*ALK*	*ALK*	*EML4-ALK*	YES	5.171	0.0007	exon15-exon20	YES
12	*ALK*	*ALK*	*EML4-ALK*	YES	4.757	0.0007	exon15-exon20	YES
13	*ALK*	*ALK*	*EML4-ALK*	YES	4.88	0.001	exon15-exon20	YES
14	*ALK*	*ALK*	*EML4-ALK*	YES	3.688	0.0007	exon15-exon20	YES
15	*ALK*	*ALK*	*EML4-ALK*	YES	5.257	0.0007	exon15-exon20	YES
16	*ALK*	*ALK*	*HIP1-ALK*	YES	5.145	0.0007	exon15-exon20	YES
17	*ALK*	*ALK*	*EML4-ALK*	YES	4.059	0.0007	exon14-exon15	YES
18	*ROS1*	*N/A*	*CD74-ROS1*	YES	*N/A*	*N/A*	*N/A*	*N/A*
19	*ROS1*	*N/A*	*EZR-ROS1*	YES	*N/A*	*N/A*	*N/A*	*N/A*
20	*ROS1*	*N/A*	*EZR-ROS1*	YES	*N/A*	*N/A*	*N/A*	*N/A*
21	*ROS1*	*N/A*	*EZR-ROS1*	YES	*N/A*	*N/A*	*N/A*	*N/A*
22	*ROS1*	*N/A*	*CD74-ROS1*	YES	*N/A*	*N/A*	*N/A*	*N/A*
23	*ROS1*	*N/A*	*CD74-ROS1*	YES	*N/A*	*N/A*	*N/A*	*N/A*
24	*ROS1*	*N/A*	*SDC4-ROS1*	YES	*N/A*	*N/A*	*N/A*	*N/A*

24 FFPE samples (NSCLC) were tested with the **Oncomine Precision Assay** on **Genexus Integrated Sequencer**.

These samples were previously tested using **FISH** and **IHC** (*ALK* only), and determined to be either *ALK* or *ROS1* fusion positive.

Verification results showed **100% agreement** when using the Oncomine Precision Assay compared to both methods,

In addition, all 17 *ALK* positive samples had concordant detection using the exon tiling imbalance method.

NOTE: ROS1 does not have exon tiling imbalance capability on Oncomine Precision Assay.

NOTE: *Testing was performed at multiple internal R&D laboratories as part of product verification testing. Samples were run at different plexy levels. Additional verification and validation testing will be performed using the Oncomine Precision Assay and FFPE samples; therefore final performance values can change.*

Complete Detection of NTRK Isoforms Using SeraCare FFPE Control

SeraSeq® FFPE NTRK Fusion RNA Reference Material	
Oncomine Precision Assay Detects All NTRK Isoforms	✓ TPM3(7) - NTRK1(10)
	✓ LMNA(11) - NTRK1(11)
	✓ IRF2BP2(1) - NTRK1(10)
	✓ SQSTM1(5) - NTRK1(10)
	✓ TFG(5) - NTRK1(10)
	✓ AFAP1(14) - NTRK2(12)
	✓ NACC2(4) - NTRK2(13)
	✓ QKI(6) - NTRK2(16)
	✓ TRIM24(12) - NTRK2(15)
	✓ PAN3(1) - NTRK2(17)
	✓ ETV6(4) - NTRK3(14)
	✓ ETV6(4) - NTRK3(15)
	✓ ETV6(5) - NTRK3(14)
	✓ ETV6(5) - NTRK3(15)
	✓ BTBD1(4) - NTRK3(14)

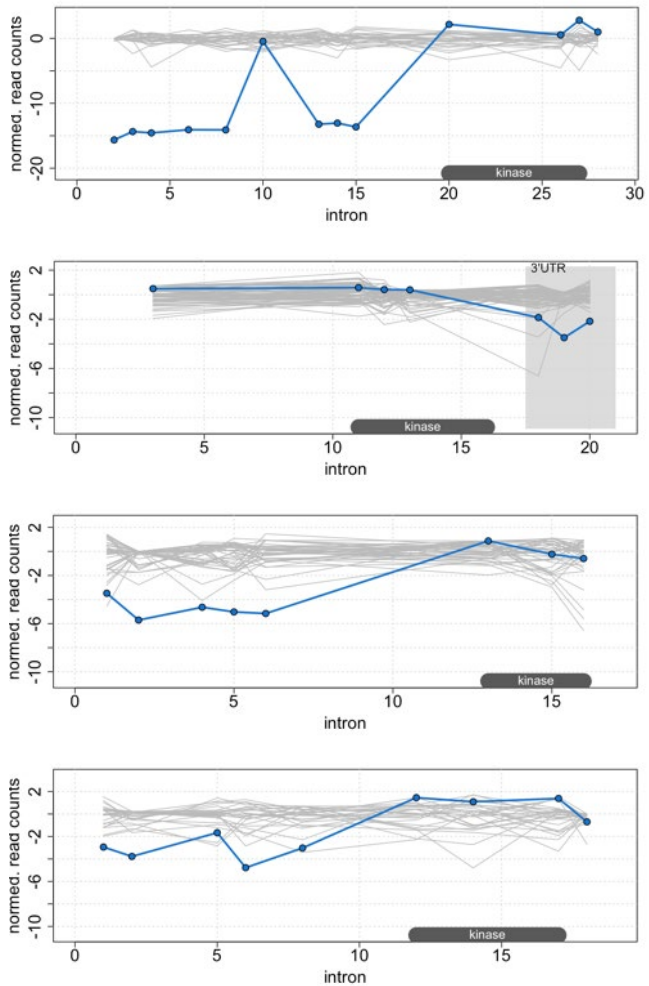
seracare™

Metric	5% Dilution
# of Runs	30
# of NTRK Fusion Isoforms	15
# of Total Datapoints	450
False Negative	4
False Positive	4
Sensitivity	**99.1%**
PPV	**100%**

- The SeraCare SeraSeq® FFPE NTRK Fusion RNA Reference Material (01710-1031) contains 15 unique NTRK isoforms (5 NTRK1, 5 NTRK2, and 5 NTRK3).
- A 5% dilution (within a normal background) of the NTRK control was made to reduce the transcript levels of the various fusion isoforms.
- The dilution was tested with multiple replicates using the **Oncomine Precision Assay** and the **Genexus Integrated Sequencer.**
- Verification results showed ability of the assay to detect all 15 NTRK fusions using the targeted isoform designs and measuring the read counts for each isoform.
- Replicate testing demonstrated high sensitivity and specificity at 5% dilutions above 33 fusion reads.

NOTE: *Testing was performed at multiple internal R&D laboratories as part of product verification testing. Samples were run at different plexy levels, resulting in an average mapped RNA reads of 190,972 for the 10% dilution dataset and an average mapped RNA reads of 691,064 for the 5% dilution dataset. Additional verification and validation testing will be performed using the Oncomine Precision Assay and SeraCare NTRK control; therefore final performance values can change.*

Detection of *ALK*, *FGFR3*, *NTRK1*, and *RET* Fusions Using Exon Tiling Imbalance



Fusion driver	Sample	Predicted breakpoint range	Imbalance score	P-value
ALK	Tri-Fusion Cell Line Mixture with *ALK*, *RET*, and *ROS1* fusions	ex15-ex20	4.74	7e-04
FGFR3	RT4 cell line	ex13-3pUTR	1.74	3.9e-03
NTRK1	KM12 cell line	ex6-ex13	2.58	4.3e-03
RET	Tri-Fusion Cell Line Mixture with *ALK*, *RET*, and *ROS1* fusions	ex8-ex12	2.12	3e-03

Performance of Genexus and Oncomine Comprehensive Assay v3

	SNV/INDEL	CNV	Fusions
Sensitivity	98.3%	97.2%	97.6%
PPV	99.0%	N/A	99.6%
Inter-run reproducibility	98.6%	97.8%	95.7%
Intra-run reproducibility	98.6%	96.7%	95.9%

SNV and INDEL performance was assessed using Acrometrix Hotspot Control and SeraCare Seraseq Tri-Level Mutation DNA Mix
CNV performance was assessed using SeraCare CNV controls
Fusion performance was assessed using Horizon HD789 and SeraCare Fusion Mix v3, Horizon HD784 ALK*, and LC/2 ad cell line*

A New world of NGS Means a New Day for your Lab

Now is the time to start with NGS with…

- One day Ion Torrent Genexus workflow*
- Complete automation from specimen to report and only 10 min hands on time*
- Oncomine Precision Assay
- No need for batching
- First in class implementation support

..and get key 50 gene NGS profile result in the same time as your IHC for comprehensive profile

*Specimen-to-report workflow will be available after the Ion Torrent™ Genexus™ Purification System and integrated reporting capabilities are added in 2020.
The content provided herein may relate to products that have not been fully validated by Thermo Fisher Scientific and is subject to change without notice.

*The **Ion Torrent™ Genexus™** is currently Research Use Only, however*

"Thermo Fisher Scientific intent to seek regulatory marketing authorization of the system so that it can be potentially made available in every clinical setting.

Additionally we also plan to develop and seek approval for a broad portfolio of diagnostic assays in oncology.

*The **Ion Torrent™ Genexus™** has been developed with the intent to shift the cancer testing paradigm in the future, that is what we worked for and will keep on working for".*

Thank you
Q&A

ThermoFisher SCIENTIFIC

Thermo Fisher SCIENTIFIC

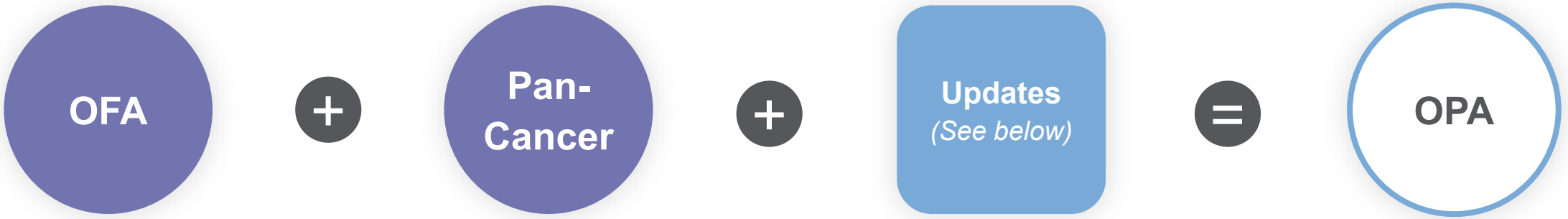
Kit Configuration, Throughput and Pricing

ThermoFisher SCIENTIFIC

Oncomine Precision Assay Throughput on Genexus

	Assay	1 Lane	2 Lanes	3 Lanes	4 Lanes
Max Number of Samples	Oncomine Precision Assay (for DNA & RNA workflow)	4	8	12	16
	Oncomine Precision Assay (for cfTNA workflow)	1	2	3	4

OPA Content – Progression on Other Oncomine Assays



Swap genes, emphasis on potential for future targeted therapies research

Examples:
NRG1 fusions
PTEN loss
AKT2/3 mutations

Increased fusion isoforms and exon tiling imbalance

NTRK isoforms **31** (OFA)→**150** (OPA)

Imbalance: *ALK, FGFR1, FGFR2, FGFR3, NTRK1, NTRK2 NTRK3, RET*

Resistance mutations

ALK, AR, EGFR, ERBB2, ERBB3, ESR1, FGFR1, FGFR2, FLT3, IDH1, IDH2, KIT, MAP2K1, MAP2K2, MET, NTRK1, NTRK2, NTRK3, PDGFRA, RET, ROS1, SMO

Assay Roadmap On Genexus

At Launch	January 2020			2020 and beyond
Custom Ion AmpliSeq HD Panels	Ion Torrent™ Oncomine™ **Precision** Assay (New)	Ion Torrent™ Oncomine™ **Comprehensive** Assay v3	Ion Torrent™ Oncomine™ **TCR Beta-LR** Assay	Oncomine Myeloid Research Assay
Custom Ion AmpliSeq Panels				Oncomine *BRCA* Research Assay
Ion AmpliSeq On-Demand Panels	Single assay to test common biomarkers from FFPE tissue and liquid biopsy samples	Testing of common and rare biomarkers Clinical trials	Translational and clinical research Immuno-oncology research	***More assays to come…***

Service and Support

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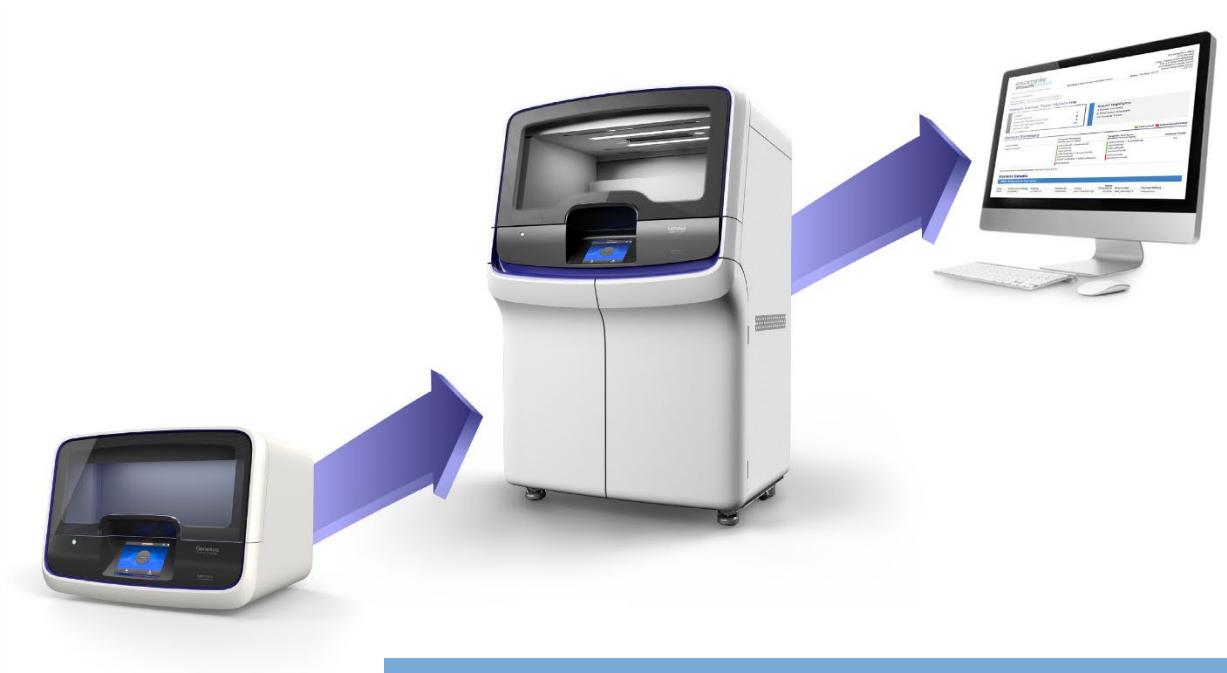

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First Experience from the “New World”

The Pathologist - Webinar

5th December 2019

José Luis Costa

(jcosta@ipatimup.pt)

INSTITUTO DE INVESTIGAÇÃO E INOVAÇÃO EM SAÚDE UNIVERSIDADE DO PORTO

ipatimup

Ipatimup – Porto - Portugal

- Biggest research in health institute in Portugal (1250 researchers);
- Cancer, Neurosciences and Host-Pathogen interactions research lines

- Founding member of i3S
- Leading cancer research institute in Portugal
- Founding partner of **Porto Comprehensive Cancer Center**

NGS clinical research timeline….

Ion PGM installation

BRCA1/2 panel (77 amplicons)

RASopathies (123 amplicons)

Ampliseq Fusion panel
Ampliseq Colon & Lung panel

Oncomine cfDNA lung assay

2011 | 2012 | 2013 | 2014 | 2015 | 2016 | 2017 | 2018 | 2019


Ion OT2


Ion Chef


Ion Chef


Ion Chef


Ion PGM

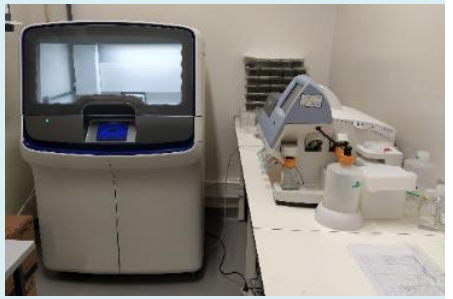

Ion Proton

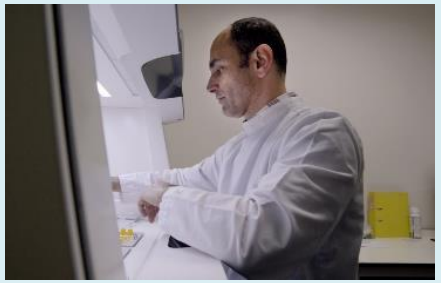

Ion S5XL


Ion S5


Ion Reporter

Genexus System





First Experience from IPATIMUP, Portugal

Commercial control samples

- Horizon and SeraCare FFPE and cfDNA reference material

Clinical research samples

- Lung cancer tissue and liquid biopsy research samples
- Previously characterize on an Ion S5XL system

First Experience from IPATIMUP, Portugal

Genexus Software | Ion Torrent

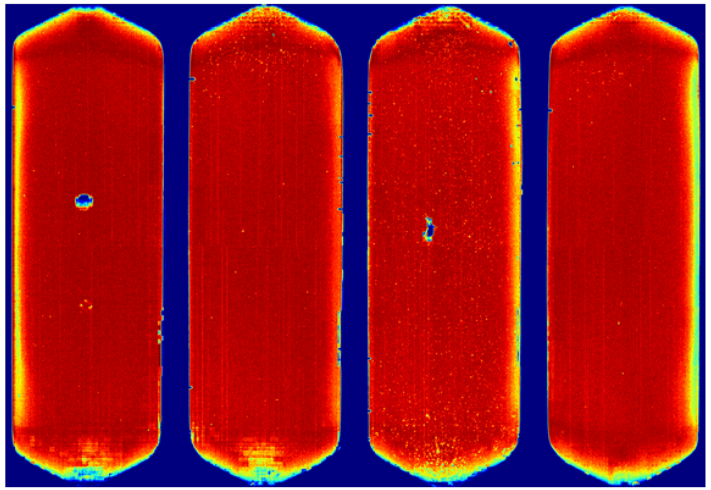
Samples Runs Monitor Results

Results > Run Results > View Results

OPA - GX5 - Liquid Biopsy TN...

Assay: OPA - GX5 - Liquid Biopsy TNA - EA2-20190827
Run Plan : Run02_27AUG19_OPA_IPEAPh2

Loading Density



89.21%	5.84G	58,534,613	99.06%
Loading	Total Bases	Final Reads	Mean Raw Accuracy 1x

Loading 89.21% — 120,856,893
Wells with ISPs / Total Addressable Wells
120,856,893 / 135,481,643

Empty Wells 10.79%

Enrichment 99.29% — 119,996,579
Wells with Live ISPs / Wells with ISPs
119,996,579 / 120,856,893

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Wells with Control ISPs 0.01%
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Low Quality 18.25%
21,901,337 / 119,985,204

Adapter Dimer 0.02%
29,212 / 119,985,204

Run Samples

Sample Name	Nucleic Acid Type	Barcode	Total Reads	Mean Read Length	>= Q20 Bases	Uniformity	Read Length Histogram
cfTNA_1_27AUG19_IPEAPh2	TNA	IonHDdual_0125	13,150,825	101 bp	1,150,409,567	Not Available	0 50 100 150 200 250 300
cfTNA_2_27AUG19_IPEAPh2	TNA	IonHDdual_0126	13,700,575	100 bp	1,192,236,490	Not Available	0 50 100 150 200 250 300
cfTNA_3_27AUG19_IPEAPh2	TNA	IonHDdual_0127	16,994,843	96 bp	1,420,277,918	Not Available	0 50 100 150 200 250 300

First Experience from IPATIMUP, Portugal

Genexus Software | Ion Torrent

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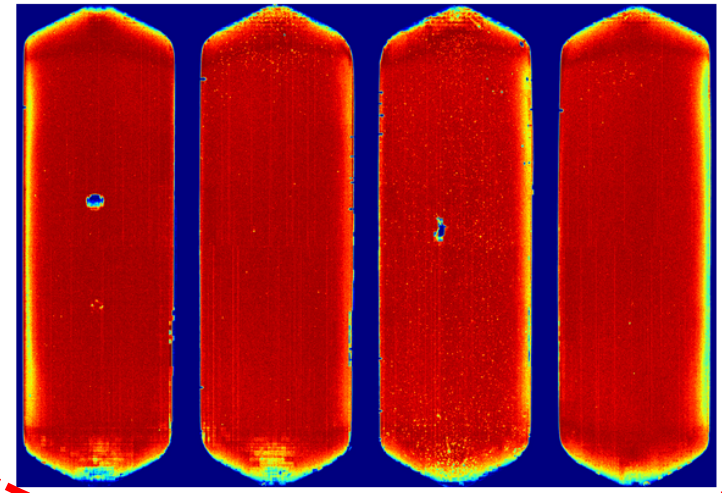

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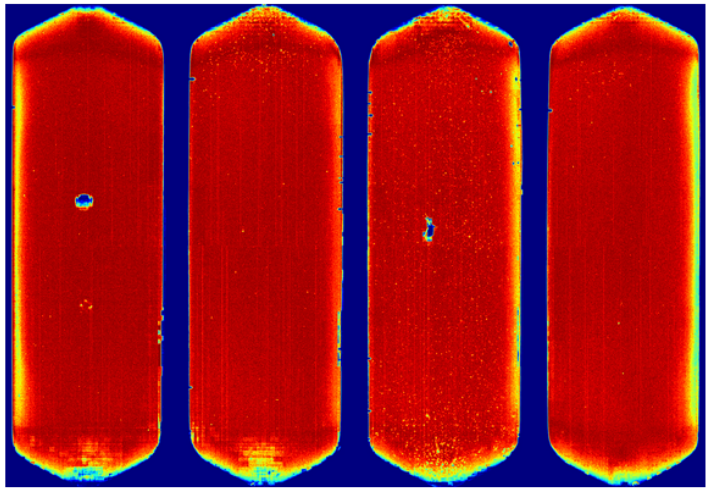

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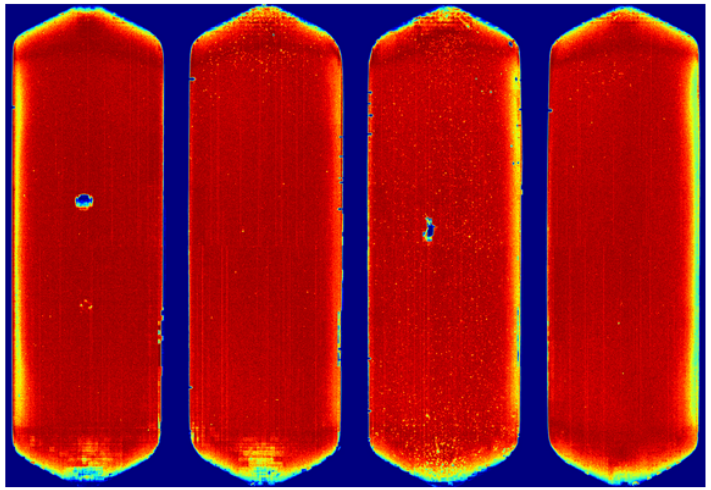

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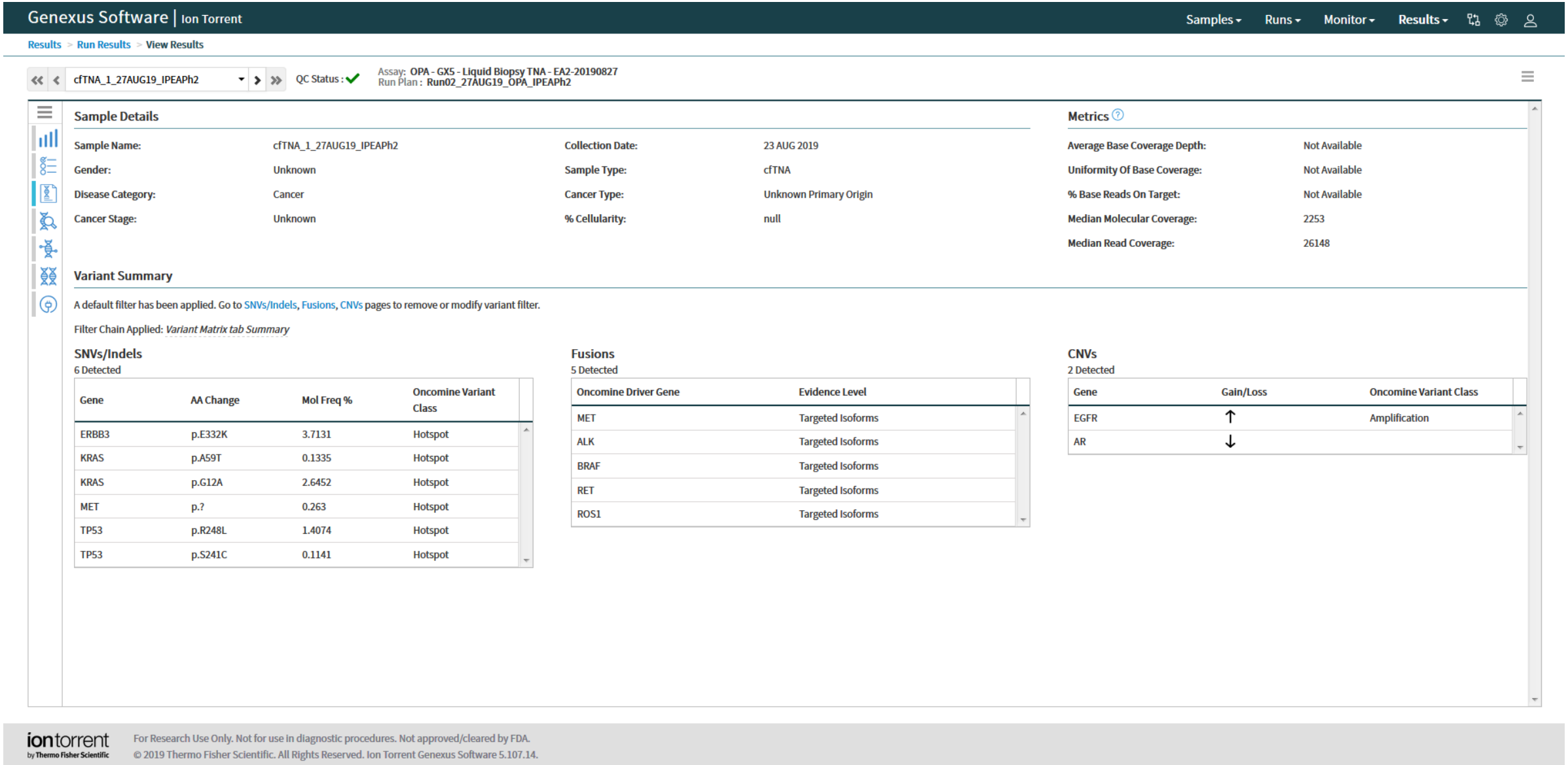
Sample Name	Nucleic Acid Type	Barcode	Total Reads	Mean Read Length	>= Q20 Bases	Uniformity	Read Length Histogram
cfTNA_1_27AUG19_IPEAPh2	TNA	IonHDdual_0125	13,150,825	101 bp	1,150,409,567	Not Available	0 50 100 150 200 250 300
cfTNA_2_27AUG19_IPEAPh2	TNA	IonHDdual_0126	13,700,575	100 bp	1,192,236,490	Not Available	0 50 100 150 200 250 300
cfTNA_3_27AUG19_IPEAPh2	TNA	IonHDdual_0127	16,994,843	96 bp	1,420,277,918	Not Available	0 50 100 150 200 250 300

First Experience from IPATIMUP, Portugal



Genexus Software | Ion Torrent

Samples Runs Monitor Results

Results > Run Results > View Results

cfTNA_1_27AUG19_IPEAPh2 QC Status : ✔

Assay: OPA - GX5 - Liquid Biopsy TNA - EA2-20190827
Run Plan : Run02_27AUG19_OPA_IPEAPh2

Sample Details

Sample Name:	cfTNA_1_27AUG19_IPEAPh2	Collection Date:	23 AUG 2019
Gender:	Unknown	Sample Type:	cfTNA
Disease Category:	Cancer	Cancer Type:	Unknown Primary Origin
Cancer Stage:	Unknown	% Cellularity:	null

Metrics

Average Base Coverage Depth:	Not Available
Uniformity Of Base Coverage:	Not Available
% Base Reads On Target:	Not Available
Median Molecular Coverage:	2253
Median Read Coverage:	26148

Variant Summary

A default filter has been applied. Go to SNVs/Indels, Fusions, CNVs pages to remove or modify variant filter.

Filter Chain Applied: *Variant Matrix tab Summary*

SNVs/Indels

6 Detected

Gene	AA Change	Mol Freq %	Oncomine Variant Class
ERBB3	p.E332K	3.7131	Hotspot
KRAS	p.A59T	0.1335	Hotspot
KRAS	p.G12A	2.6452	Hotspot
MET	p.?	0.263	Hotspot
TP53	p.R248L	1.4074	Hotspot
TP53	p.S241C	0.1141	Hotspot

Fusions

5 Detected

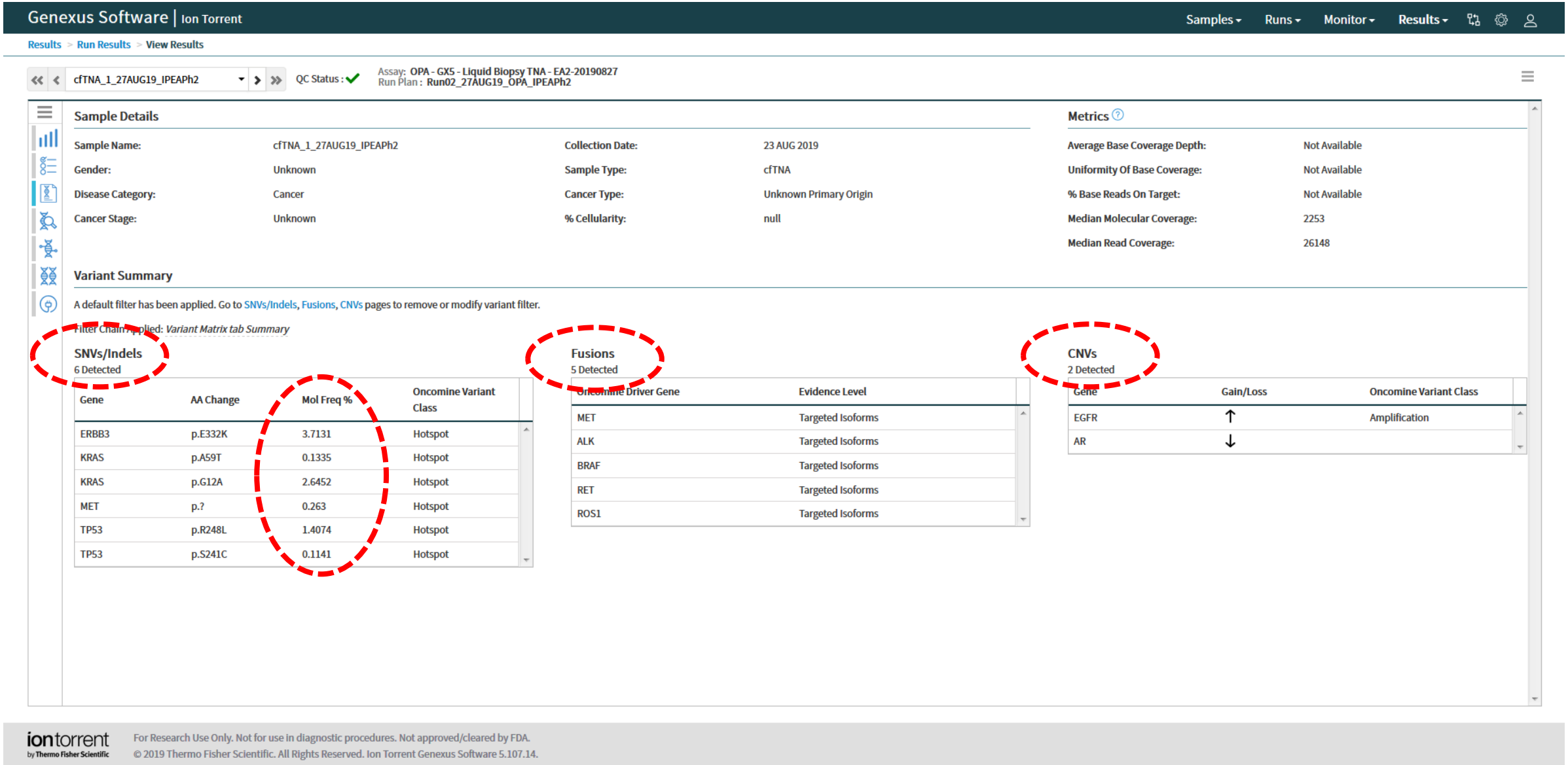
Oncomine Driver Gene	Evidence Level
MET	Targeted Isoforms
ALK	Targeted Isoforms
BRAF	Targeted Isoforms
RET	Targeted Isoforms
ROS1	Targeted Isoforms

CNVs

2 Detected

Gene	Gain/Loss	Oncomine Variant Class
EGFR	↑	Amplification
AR	↓	

First Experience from IPATIMUP, Portugal



Genexus Software | Ion Torrent

Samples Runs Monitor Results

Results > Run Results > View Results

cfTNA_1_27AUG19_IPEAPh2 QC Status : ✔

Assay: OPA - GX5 - Liquid Biopsy TNA - EA2-20190827
Run Plan : Run02_27AUG19_OPA_IPEAPh2

Sample Details

Sample Name:	cfTNA_1_27AUG19_IPEAPh2	Collection Date:	23 AUG 2019
Gender:	Unknown	Sample Type:	cfTNA
Disease Category:	Cancer	Cancer Type:	Unknown Primary Origin
Cancer Stage:	Unknown	% Cellularity:	null

Metrics

Average Base Coverage Depth:	Not Available
Uniformity Of Base Coverage:	Not Available
% Base Reads On Target:	Not Available
Median Molecular Coverage:	2253
Median Read Coverage:	26148

Variant Summary

A default filter has been applied. Go to SNVs/Indels, Fusions, CNVs pages to remove or modify variant filter.

Filter Chain Applied: *Variant Matrix tab Summary*

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KRAS	p.G12A	2.6452	Hotspot
MET	p.?	0.263	Hotspot
TP53	p.R248L	1.4074	Hotspot
TP53	p.S241C	0.1141	Hotspot

Fusions

5 Detected

Oncomine Driver Gene	Evidence Level
MET	Targeted Isoforms
ALK	Targeted Isoforms
BRAF	Targeted Isoforms
RET	Targeted Isoforms
ROS1	Targeted Isoforms

CNVs

2 Detected

Gene	Gain/Loss	Oncomine Variant Class
EGFR	↑	Amplification
AR	↓	

First Experience from IPATIMUP, Portugal

SNVs/InDels

Sample ID	Gene	Variant	Molecular frequency (%)	
			Run 1	Run 2
FFPE1	AKT1	p.?	4,2	4,9
FFPE1	BRAF	p.V600E	15,5	19,3
FFPE1	CDKN2A	p.R58*	nd	9,3
FFPE1	CTNNB1	p.S33Y	5,9	6,2
FFPE1	CTNNB1	p.S45del	4,8	4,3
FFPE1	EGFR	p.A767_V769dup	3,5	5,1
FFPE1	EGFR	p.E746_A750del	3,1	nd
FFPE1	EGFR	p.G719S	7,4	7,1
FFPE1	GNA11	p.Q209L	3,9	2,7
FFPE1	KRAS	p.G13D	2,8	3,7
FFPE1	MAP2K1	p.Q56P	3,8	3,4
FFPE1	PIK3CA	p.E545K	3,8	3,5
FFPE1	PIK3CA	p.H1047R	19,8	17,6
FFPE1	TP53	p.S241C	9,0	5,8
FFPE1	TP53	p.S241F	9,0	10,2
FFPE2	AKT3	p.Q78K	nd	2,2
FFPE2	BRAF	p.V600E	64,3	69,7
FFPE2	EGFR	p.E746_A750del	3,5	3,6
FFPE2	EGFR	p.G719S	5,4	6,3
FFPE2	EGFR	p.L858R	3,9	5,1
FFPE2	EGFR	p.L861Q	3,9	3,4
FFPE2	EGFR	p.T790M	4,9	4,1
FFPE2	PIK3CA	p.H1047R	50,8	54,6
FFPE3	CTNNB1	p.S33Y	51,6	51,0
FFPE3	EGFR	p.G719S	31,5	32,6
FFPE3	KRAS	p.A146T	3,8	3,7
FFPE3	KRAS	p.G12D	5,9	4,4
FFPE3	KRAS	p.G13D	4,1	4,0
FFPE3	KRAS	p.Q61H	3,9	3,1
FFPE3	MAP2K1	p.Q56P	49,4	52,2
FFPE3	NRAS	p.G12V	4,0	6,0
FFPE3	NRAS	p.Q61K	3,6	3,8
FFPE5	BRAF	p.V600E	15,3	14,3
FFPE5	EGFR	p.E746_A750del	13,1	10,8
FFPE5	EGFR	p.L858R	14,6	16,1
FFPE5	ERBB2	p.Y772_A775dup	15,9	14,8
FFPE5	IDH1	p.R132C	14,3	13,3
FFPE5	KRAS	p.G12D	14,2	14,5
FFPE5	PIK3CA	p.E542K	16,3	16,4

Fusions

Sample ID	Driver Gene	Run 1	Run 2
FFPE2	ALK	Detected	Detected
FFPE2	BRAF	Detected	Detected
FFPE2	NTRK1	Detected	Detected
FFPE2	NTRK2	Detected	Detected
FFPE2	NTRK3	Detected	Detected
FFPE5	FGFR3	Detected	Detected

CNVs

Sample ID	Gene	Run 1	Run 2
FFPE1	CDKN2A	loss	loss
FFPE1	MET	gain	gain
FFPE5	MET	gain	gain
FFPE5	AR	loss	Loss
FFPE5	FGFR3	loss	nd

- Different variant types can be detected;

nd – not detected

First Experience from IPATIMUP, Portugal

SNVs/InDels

Sample ID	Gene	Variant	Molecular frequency (%)	
			Run 1	Run 2
FFPE1	AKT1	p.?	4,2	4,9
FFPE1	BRAF	p.V600E	15,5	19,3
FFPE1	CDKN2A	p.R58*	nd	9,3
FFPE1	CTNNB1	p.S33Y	5,9	6,2
FFPE1	CTNNB1	p.S45del	4,8	4,3
FFPE1	EGFR	p.A767_V769dup	3,5	5,1
FFPE1	EGFR	p.E746_A750del	3,1	nd
FFPE1	EGFR	p.G719S	7,4	7,1
FFPE1	GNA11	p.Q209L	3,9	2,7
FFPE1	KRAS	p.G13D	2,8	3,7
FFPE1	MAP2K1	p.Q56P	3,8	3,4
FFPE1	PIK3CA	p.E545K	3,8	3,5
FFPE1	PIK3CA	p.H1047R	19,8	17,6
FFPE1	TP53	p.S241C	9,0	5,8
FFPE1	TP53	p.S241F	9,0	10,2
FFPE2	AKT3	p.Q79K	nd	2,2
FFPE2	BRAF	p.V600E	64,3	69,7
FFPE2	EGFR	p.E746_A750del	3,5	3,6
FFPE2	EGFR	p.G719S	5,4	6,3
FFPE2	EGFR	p.L858R	3,9	5,1
FFPE2	EGFR	p.L861Q	3,9	3,4
FFPE2	EGFR	p.T790M	4,9	4,1
FFPE2	PIK3CA	p.H1047R	50,8	54,6
FFPE3	CTNNB1	p.S33Y	51,6	51,0
FFPE3	EGFR	p.G719S	31,5	32,6
FFPE3	KRAS	p.A146T	3,8	3,7
FFPE3	KRAS	p.G12D	5,9	4,4
FFPE3	KRAS	p.G13D	4,1	4,0
FFPE3	KRAS	p.Q61H	3,9	3,1
FFPE3	MAP2K1	p.Q56P	49,4	52,2
FFPE3	NRAS	p.G12V	4,0	6,0
FFPE3	NRAS	p.Q61K	3,6	3,8
FFPE5	BRAF	p.V600E	15,3	14,3
FFPE5	EGFR	p.E746_A750del	13,1	10,8
FFPE5	EGFR	p.L858R	14,6	16,1
FFPE5	ERBB2	p.Y772_A775dup	15,9	14,8
FFPE5	IDH1	p.R132C	14,3	13,3
FFPE5	KRAS	p.G12D	14,2	14,5
FFPE5	PIK3CA	p.E542K	16,3	16,4

Fusions

Sample ID	Driver Gene	Run 1	Run 2
FFPE2	ALK	Detected	Detected
FFPE2	BRAF	Detected	Detected
FFPE2	NTRK1	Detected	Detected
FFPE2	NTRK2	Detected	Detected
FFPE2	NTRK3	Detected	Detected
FFPE5	FGFR3	Detected	Detected

CNVs

Sample ID	Gene	Run 1	Run 2
FFPE1	CDKN2A	loss	loss
FFPE1	MET	gain	gain
FFPE5	MET	gain	gain
FFPE5	AR	loss	Loss
FFPE5	FGFR3	loss	nd

- Different variant types can be detected;
- Reproducibility high between runs;
- Curiously, deviation of molecular frequency is bigger for higher molecular frequencies (MF<10% SD:0.6 – MF>10% SD:1.4);

nd – not detected

First Experience from IPATIMUP, Portugal

SNVs/InDels

Sample ID	Gene	Variant	Molecular frequency (%)	
			Run 1	Run 2
FFPE1	AKT1	p.?	4,2	4,9
FFPE1	BRAF	p.V600E	15,5	19,3
FFPE1	CDKN2A	p.R58*	nd	9,3
FFPE1	CTNNB1	p.S33Y	5,9	6,2
FFPE1	CTNNB1	p.S45del	4,8	4,3
FFPE1	EGFR	p.A767_V769dup	3,5	5,1
FFPE1	EGFR	p.E746_A750del	3,1	nd
FFPE1	EGFR	p.G719S	7,4	7,1
FFPE1	GNA11	p.Q209L	3,9	2,7
FFPE1	KRAS	p.G13D	2,8	3,7
FFPE1	MAP2K1	p.Q56P	3,8	3,4
FFPE1	PIK3CA	p.E545K	3,8	3,5
FFPE1	PIK3CA	p.H1047R	19,8	17,6
FFPE1	TP53	p.S241C	9,0	5,8
FFPE1	TP53	p.S241F	9,0	10,2
FFPE2	AKT3	p.Q78K	nd	2,2
FFPE2	BRAF	p.V600E	64,3	69,7
FFPE2	EGFR	p.E746_A750del	3,5	3,6
FFPE2	EGFR	p.G719S	5,4	6,3
FFPE2	EGFR	p.L858R	3,9	5,1
FFPE2	EGFR	p.L861Q	3,9	3,4
FFPE2	EGFR	p.T790M	4,9	4,1
FFPE2	PIK3CA	p.H1047R	50,8	54,6
FFPE3	CTNNB1	p.S33Y	51,6	51,0
FFPE3	EGFR	p.G719S	31,5	32,6
FFPE3	KRAS	p.A146T	3,8	3,7
FFPE3	KRAS	p.G12D	5,9	4,4
FFPE3	KRAS	p.G13D	4,1	4,0
FFPE3	KRAS	p.Q61H	3,9	3,1
FFPE3	MAP2K1	p.Q56P	49,4	52,2
FFPE3	NRAS	p.G12V	4,0	6,0
FFPE3	NRAS	p.Q61K	3,6	3,8
FFPE5	BRAF	p.V600E	15,3	14,3
FFPE5	EGFR	p.E746_A750del	13,1	10,8
FFPE5	EGFR	p.L858R	14,6	16,1
FFPE5	ERBB2	p.Y772_A775dup	15,9	14,8
FFPE5	IDH1	p.R132C	14,3	13,3
FFPE5	KRAS	p.G12D	14,2	14,5
FFPE5	PIK3CA	p.E542K	16,3	16,4

Fusions

Sample ID	Driver Gene	Run 1	Run 2
FFPE2	ALK	Detected	Detected
FFPE2	BRAF	Detected	Detected
FFPE2	NTRK1	Detected	Detected
FFPE2	NTRK2	Detected	Detected
FFPE2	NTRK3	Detected	Detected
FFPE5	FGFR3	Detected	Detected

CNVs

Sample ID	Gene	Run 1	Run 2
FFPE1	CDKN2A	loss	loss
FFPE1	MET	gain	gain
FFPE5	MET	gain	gain
FFPE5	AR	loss	Loss
FFPE5	FGFR3	loss	nd

- Different variant types can be detected;
- Reproducibility high between runs;
- Curiously, deviation of molecular frequency is bigger for higher molecular frequencies (MF<10% SD:0.6 – MF>10% SD:1.4);

Accuracy of 98% (95%CI 94% to 99%);

Sensitivity of 96 % (95%CI 89% to 99%)

Specificity of 100% (95%CI 94% to 100%).

nd – not detected

First Experience from IPATIMUP, Portugal

Commercial control samples

- Horizon and SeraCare FFPE and cfDNA reference material

Clinical research samples

- Lung cancer tissue and liquid biopsy research samples
- Previously characterize on an Ion S5XL system

Tissue biopsies – RNA and DNA

Lung cancer FFPE sample were sequenced using Colon and Lung or Lung Fusion panel on Ion S5XL system and the Oncomine Precision Assay on Genexus instrument

Variants covered by both panels were detected in both systems at similar allelic frequencies

Additional variants were detected using the Oncomine Precision Assay

Sample 1 presented additional p.T790M mutation that had not been previously identified (intra-sample heterogeneity?)

FFPE	Gene	AA Change	Genexus	Ion S5XL
Sample 1	**EGFR**	**p.L858R**	**43,8**	**31,3**
	TP53	p.R248Q	1,8	nd
	EGFR	**p.T790M**	**1,6**	**nd**
	ALK	p.A1200V	1,5	nd
Sample 2	**EGFR**	**p.L747_P753delinsS**	**28,6**	**20,1**
	TP53	p.C176R	3,9	nd
	RET	p.G810S	1,6	nd
	EGFR	p.P848L	1,6	nd
	FGFR3	p.R399C	0,3	nd
Sample 3	**ALK**	**fusion**	**detected**	**detected**
Sample 4	**BRAF**	**p.V600E**	**47,8**	**36,3**
	TP53	p.R175C	13,8	nd
	FGFR2	p.A648T	6,9	nd
	EGFR	p.R836C	6,2	nd
	ERBB3	p.V104M	4,4	nd
	PDGFRA	p.T849C	4,2	nd
	PIK3CA	p.V344M	3,7	nd
	MAP2K1	p.K57N	2,8	nd
	GNAQ	p.R183Q	2,1	nd
	EGFR	p.V769M	1,8	nd
Sample 5	**KRAS**	**p.G12D**	**58,3**	**44,7**
	TP53	p.C176Y	31,4	nd
	FGFR3	p.R399C	3,6	nd
	CDKN2A	p.R58Q	3,1	nd
	RET	p.R912Q	2,5	nd
	BRAF	p.D594N	2,3	nd

nd – not detected

Liquid biopsies – Lung cancer plasma sample at recurrence

9AM
10min
hands on

Plasma collection

11AM
90min
hands on

cfTNA isolation

4PM
10min
hands on

Variant detection

9AM
10min
hands on

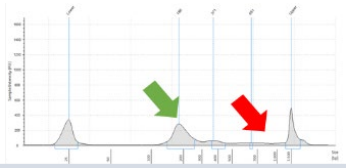
Report

1. Plasma collection

BD Vacutainer PPT
(K2EDTA)

2. cfTNA isolation

MagMax cfTNA
extraction kit



3. NGS variant identification

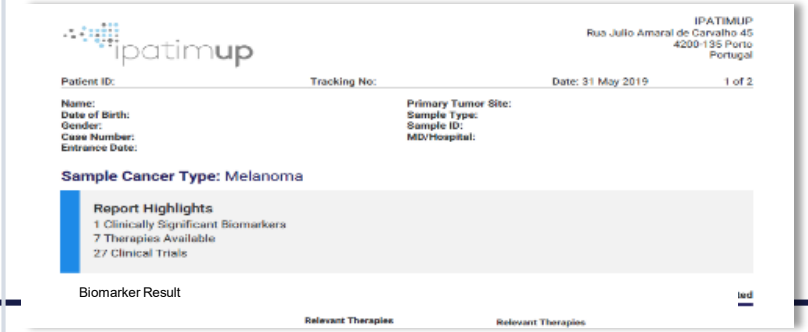
Genexus

\+

Oncomine
Precision Assay

Gene	AA Change	Mol. Freq. %
EGFR	p.E746_A750del	1,59
EGFR	p.T790M	1,78

4. OKR Report


ipatimup

IPATIMUP
Rua Júlio Amaral de Carvalho 45
4200-135 Porto
Portugal

Patient ID: Tracking No: Date: 31 May 2019 1 of 2

Name:
Date of Birth:
Gender:
Case Number:
Entrance Date:

Primary Tumor Site:
Sample Type:
Sample ID:
MD/Hospital:

Sample Cancer Type: Melanoma

Report Highlights
1 Clinically Significant Biomarkers
7 Therapies Available
27 Clinical Trials

Biomarker Result

Relevant Therapies Relevant Therapies

- Variant Result
- Interpretation

Sample

< 24h

Report

ThermoFisher SCIENTIFIC

Genexus implementation scenarios

Stand-alone

All-in-one

Genexus

High volume
Routine assays

Occasional assays
Short TAT assays

Genexus implementation scenarios

Stand-alone

All-in-one

Genexus

High volume
Routine assays

Occasional assays
Short TAT assays

Bundle

our strategy

Workhorse

Ion Chef

Ion S5XL

High volume
Routine assays

Fast track

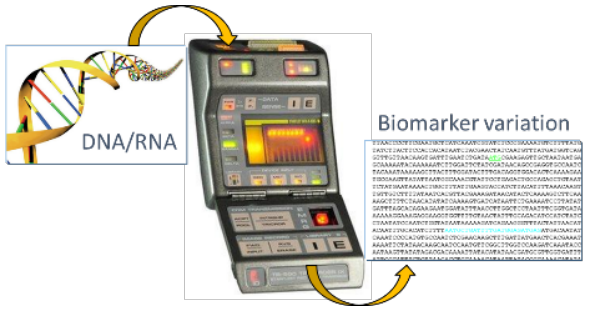
Genexus

Occasional assays
Short TAT assays

Overall impression

- ALL-IN-ONE solution from nucleic acids to variants;
- ACCESSIBLE: no *a priori* NGS expertize needed;
- SIMPLE: all I needed was a pipette to add my samples to be sequenced;
- ROBUST: vision-system checks all your steps;
- FAST: allowed a turn around time of 24h from plasma to report.

This system allows a wider implementation of NGS for genomic profiling, potentially bringing precision medicine closer to clinical practice.



Joana Reis
Sandra Coelho
José Carlos Machado

Venceslau Hespanhol
Gabriela Fernandes
Fatima Carneiro
Conceição Souto Moura

Sohaib Qureshi
Jussi Vanhatalo
Ian Grinsell
Rosella Petraroli
Andy Felton

Funding

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