ThermoFisher SCIENTIFIC

# Genexus System news

## Issue 10: May 2023

## Welcome to our next issue of Genexus System news, the first in 2023

On March 30, we held our 5th **Oncomine World virtual educational event**, so if you didn't have a chance yet, you can now see it all on demand here. You will hear many talks from your peers discussing the next-generation sequencing (NGS) contribution to precision oncology, both for research and routine patient testing. Topics include:

- The genomic instability metric (GIM) for homologous recombination deficiency (HRD) assessment, which is now available on the Ion Torrent™ Oncomine™ Comprehensive Assay Plus on the Ion GeneStudio™ system, and will be available later this year on the Ion Torrent™ Genexus™ System
- The introduction of a rapid lung NGS program in leading cancer centers
- Liquid biopsy testing, considered to be the future in precision oncology—but how far in the future?
- First presentations by Oncomine Clinical Research Grant awardees

You can also visit updated labs including hot biomarker, comprehensive genomic profiling (CGP), and Genexus System labs, as well as the Oncomine Clinical Research Grant hall.

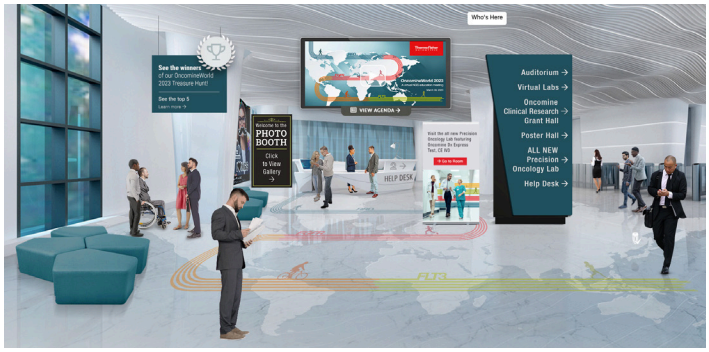
Here we will introduce two talks showcasing the use of Ion Torrent™ Oncomine™ assays on the Genexus System:

### 1. Simultaneous DNA and RNA variant testing by the Oncomine™ Precision Assay Presented

by Annarita Destro, PhD, Head of Molecular Pathology Department, Humanitas Research Hospital, Italy

In her presentation, Dr. Destro talked about her lab's extensive experience with the Oncomine Precision Assay on the Genexus System, and focused in detail on a cohort of 452 samples, where they performed simultaneous analysis of the DNA- and RNA-based variants. This has been only in recent years enabled by NGS, and still there is a debate about when it is indicated and cost-effective, compared to consequent analysis of DNA and then RNA. Based on their experience, Dr. Destro concludes that the simultaneous analysis yields more accurate results and is cost-effective.

That conclusion is based on a reduction of the amount of samples used for analysis and of the number of incompleted




analyses due to insufficient math, reduction in turn-around time (TAT) and hands-on time, as well as higher diagnostic yield. See Table 1.

**Table 1. Comparison of named variants detection yield and TAT using conventional techniques and the Oncomine Precision Assay.**

| | Conventional technique N=397 | Oncomine Precision Assay N=388 | |
|---|---|---|---|
| *EGFR* mutations | 45 **(11.3%)** | 65 **(16.7%)** | **p = 0.031** |
| Failed DNA | 2 (0.5%) | 1 (0.2%) | |
| | **Conventional technique N=397** | **Oncomine Precision Assay N=388** | |
| *ALK* translocation | 15 (3.7%) | 12 (3.1%) | |
| *ROS1* translocation | 3 (0.8%) | 3 (0.8%) | |
| *RET* translocation | 6 (1.5%) | 4 (1.0%) | |
| *MET* exon skipping | 13 (3.3%) | 19 (4.9%) | |
| Failed RNA | 5 (1.3%) | 8 (2%) | |
| Failed RNA for insufficient material | 8 **(1.8%)** | **0** | **p = 0.0076** |
| | **Conventional technique** | **Oncomine Precision Assay** | |
| Days for analysis* | **13.6** | **8.6** | |

* Enclosed days for histological assessment.

### 2. Evaluation of the Ion Torrent™ Oncomine™ Comprehensive Assay v3 on the Genexus System Presented

by Dr. Bin Wang, Senior Hospital Scientist, SydPath—St. Vincent's Hospital, Sydney, Australia

Table 2. Lower limit of detection assessment with sensitivity of 96% when Variant Allelic Frequency 3.5%–4.5%.

| Mutation | Variant type | Allelic frequency (%) | HD789A | HD789B | HD789C | HD789D | HD789E |
|---|---|---|---|---|---|---|---|
| *GNA11* Q209L | SNV high GC | 4.4 | Y | Y | Y | Y | Y |
| *AKT1* E17K | SNV high GC | 3.5 | Y | Y | N | Y | Y |
| *PIK3CA* E45K | SNV low GC | 4.5 | Y | Y | Y | Y | Y |
| *EGFR* V769_D77insASV | Long insertion | 4.4 | Y | Y | Y | Y | Y |
| *EGFR* ΔE746–A750 | Long deletion | 4.4 | Y | Y | Y | Y | Y |

Dr. Wang presented data on complete analytical evaluation of 83 samples across 16 different tumor types, which included precision, analytical specificity, and sensitivity, and limit of detection (LOD) establishment. In his summary, he highlighted complete automation of library preparation (including cDNA synthesis), template preparation, sequencing, primary data analysis, and variant reporting, as well as performance reliability and satisfactory sensitivity and specificity in SNVs, indels, CNVs, and fusion detection with detection limitation of >5% VAF for SNVs and indels, and >5 copies for CNVs.

## New Tech Note: Comparison of hands-on time in (NGS) workflows for the Genexus Integrated Sequencer and the Illumina MiSeq System

In other news, we want to introduce to you a new tech note, describing an internal activity-based cost analysis conducted by researchers at Thermo Fisher Scientific. They compared the labor costs and time required to prepare sequencing libraries and perform NGS on the Ion Torrent™ Genexus™ Integrated Sequencer and the Illumina™ MiSeq™ System using a two-pool targeted NGS panel.

As high demands for expertise and hands-on technician work and associated costs are among the biggest obstacles to widespread use of NGS, we wanted to show that NGS is considerably easier and also cheaper with the Genexus sequencer.

In this internal study, the workflow for the MiSeq System with manual library preparation took approximately 2.5 hours more hands-on time on average than the workflow for the Genexus sequencer (Thermo Fisher internal data on file). This means laboratories could get back an average of ~2.5 hours per NGS run per operator, or about 33% of an 8-hour day, by switching to the Genexus Integrated Sequencer (see Figure 1). Users of the Genexus sequencer can be more productive than users of the MiSeq System and still have time to perform other laboratory tasks. The cost of labor for an average run with 8 samples on the Genexus Integrated Sequencer was just over $7 in this study, while an equivalent run on the MiSeq System cost nearly $96.

**Read the whole tech note >>**

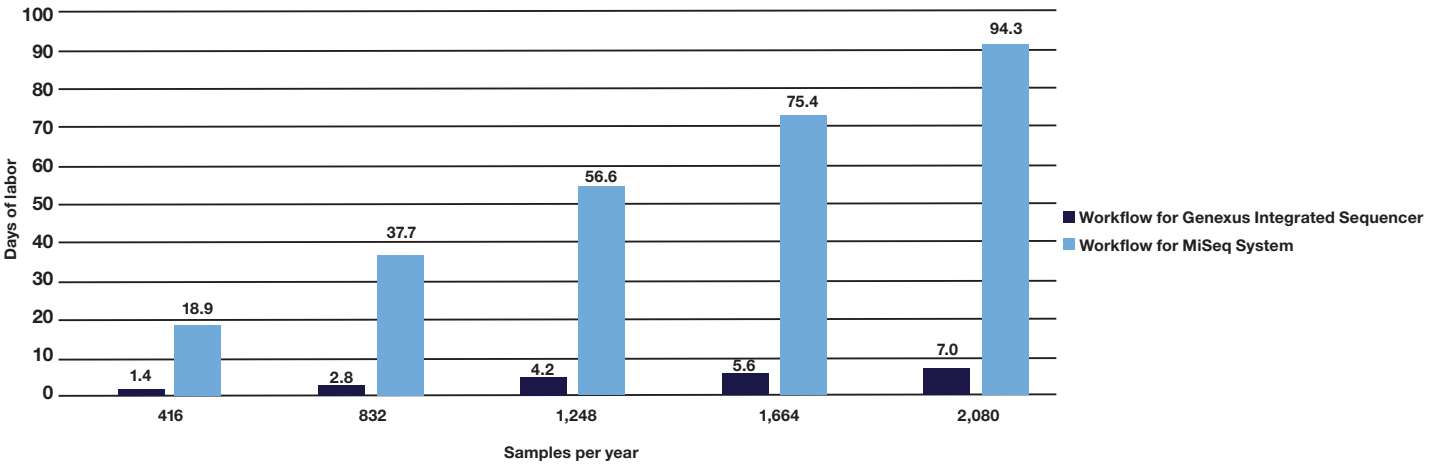
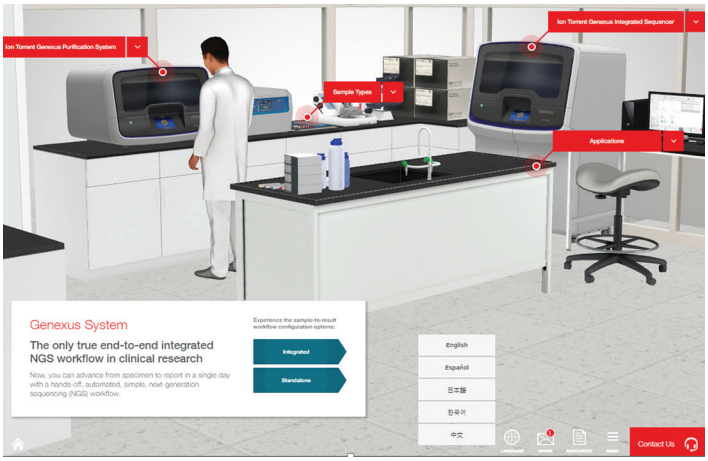



**Figure 1. Days of labor (HOT) required for an operator to prepare and sequence nucleic acid samples using the workflows for the Genexus and MiSeq systems.** Preparing and sequencing libraries for 8 samples took 13 minutes on average using the Genexus sequencer. HOT for the MiSeq System was calculated assuming a full-time employee (FTE) would take 2 hours and 54 minutes to prepare 2-pool Ion AmpliSeq™ libraries for 8 samples per run and perform 1 to 5 NGS runs per week for a total of 416 to 2,080 samples.




## Explore automating the NGS workflow from your home or office in new languages

Take a virtual tour to learn how the Genexus System simplifies NGS with walkaway automation from the comfort of your favorite chair with this newly updated 3D online demo. Engage and interact with each step of the workflow, including automated nucleic acid purification and quantification, automated NGS library preparation and sequencing, and bioinformatics analysis and reporting.

The Genexus System virtual demo is now available in Spanish, Japanese, Korean, and Mandarin Chinese, in addition to English.

**Take the virtual tour >>**

## Saving the environment

### Recyle your used Ion Torrent chips

The Ion Chip Recycling Program provides customers with a convenient way to recycle used Ion Torrent™ chips. We have partnered with reputable, certified recyclers in the United States, Canada, and Europe to collect and process the chips. To recycle your used Ion Torrent chips, simply mail your used chips to the location closest to you.

**Learn more >>**

## New on the Oncomine blog—Leveraging rapid NGS to advance hemato-oncology research

Cecilia Yeung, MD
Medical Director
Fred Hutch CLIA Laboratories

In a workshop at the Association for Molecular Pathology (AMP) annual meeting, Dr. Cecilia Yeung shared how her lab is implementing rapid NGS to advance hemato-oncology research. Read our latest blog to learn more about her lab's exciting collaboration with the National Cancer Institute (NCI) and see the data, including FLT3-ITD performance, from their analytical validation of the Ion Torrent™ Oncomine™ Myeloid Assay on the Genexus System.

**Read the blog >>**


Read more about the Genexus System at
**thermofisher.com/oncomine** and **thermofisher.com/genexus**