



Multiple Ion Torrent™ Genexus™ Systems supporting validation programs.

In this issue, we bring you a number of new stories, testimonials, and data from Genexus System users. But first, we want to share that we are working hard on making the Genexus System, and a number of next-generation sequencing (NGS) assays, compliant with the new *In Vitro* Diagnostic Regulation (IVDR) (EU) 2017/746. The Genexus System and Ion Torrent™ Oncomine™ Dx Target Test are expected to comply with CE mark requirements in 2022 with other assays to follow. Clinical laboratories rely on us to deliver validated and reliable solutions. Currently, the Oncomine Dx Target Test validation program is well underway at a number of our facilities including that pictured above. Our scientists are working around the clock, completing hundreds of experiments using >20 Genexus systems simultaneously to provide the analytical and clinical performance data needed to meet the more stringent regulation requirements for clinical evidence.

Genexus System: The true end-to-end NGS workflow

While other NGS systems claim “end to end” or “automated” workflow, the Genexus System and Ion Torrent™ assay menu provide the only true end-to-end solution on the market and attested by our customers.

The Genexus System integrates and automates nucleic acid extraction, purification, and quantitation, as well as library preparation, sequencing, analysis, and reporting under a single software ecosystem. This reduces the number of instruments and consumables required, and frees up your time for more technical applications, helping boost your lab's overall efficiency (Figure 1). With just 20 minutes of hands-on time and two touchpoints, all users can get up and running quickly with significantly less training, making NGS accessible even if your lab is new to the technology.



Quote of the month

“We can do pretty much a full week workload of our oncology NGS pipeline in the Ion Torrent™ Genexus™ Integrated Sequencer and have it automate the entire process.”

– David Seidman, PhD, MB (ASCP) ^{CM} Scientific Director for the Molecular Diagnostics and Serology Laboratories Sentara Healthcare Laboratory Services

For European laboratories who want to learn more about our IVDR transition strategy, visit thermofisher.com/genexus-IVDR

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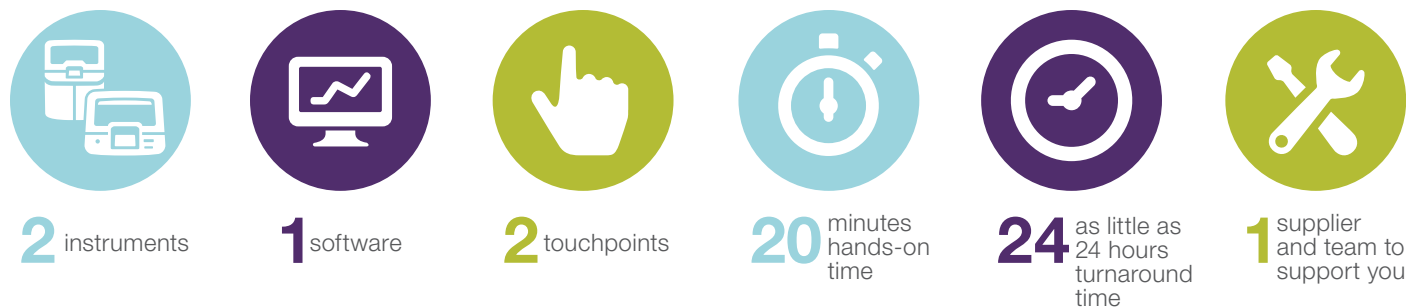
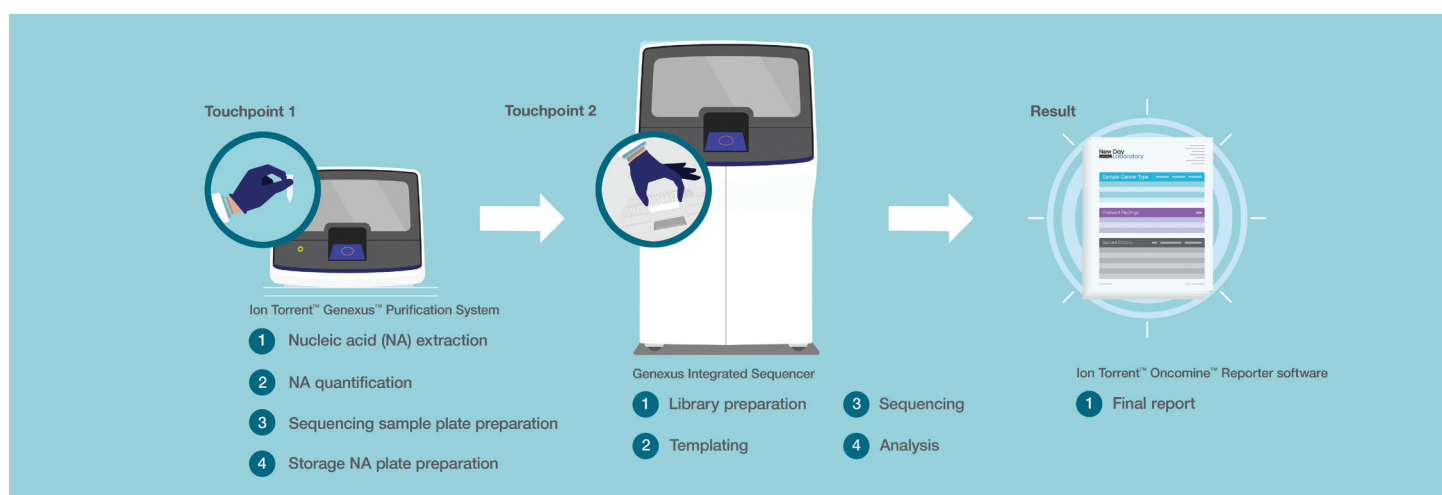


Figure 1. The Genexus System workflow makes it simple for any lab to integrate NGS.

Empower your lab to deliver NGS genomic profiles with the speed and simplicity of PCR or immunohistochemistry. The Genexus System is designed to deliver a full oncology research biomarker report in as little as one day, even for small samples and low-level variants.

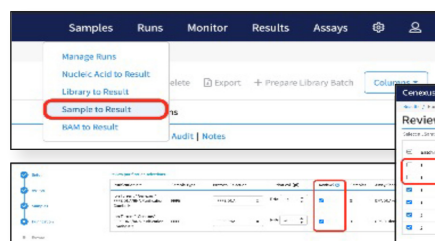
The Genexus System workflow



The Genexus Software

As shown in Figure 2, with the Ion Torrent™ Genexus Software in integrated mode samples are entered only once and tracked throughout the process to final report.

Step 1: Plan sample-to-result run



Step 2: Review purification result and select samples for sequencing



Step 3: Continue run plan to sequence selected samples



Step 4: Review run results

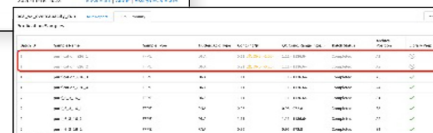


Figure 2. Screenshots of the Ion Torrent Genexus System demonstrate the complete NGS workflow, from DNA extraction to variants results and report.

Learn more about the Genexus System and your peers' experience at oncomine.com/genexus-oncology

An experience with the Genexus System: A streamlined workflow for solid tumor FFPE analysis



David Seidman, PhD, MB (ASCP) ^{CM}

Scientific Director for the Molecular Diagnostics and Serology Laboratories Sentara Healthcare Laboratory Services

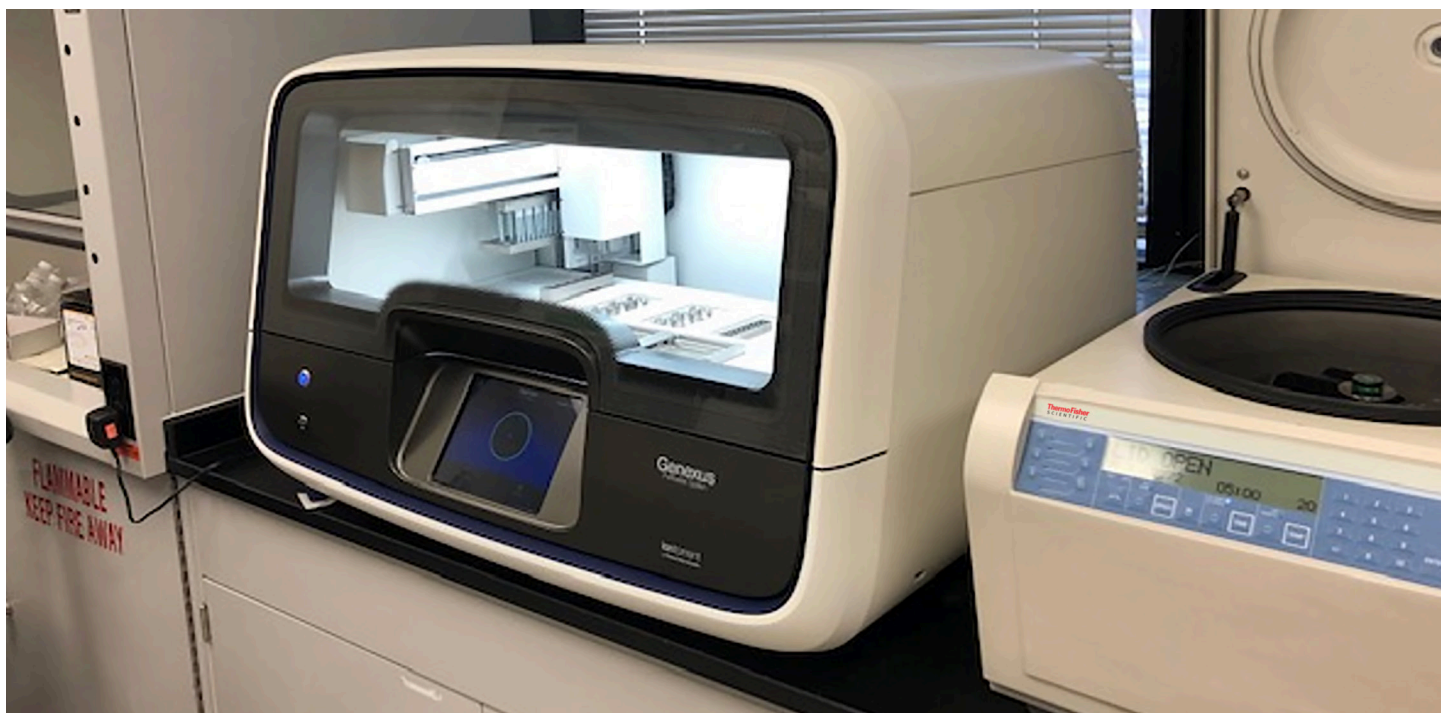
Dr. Seidman presented at Ion World 2021 on October 13, 2021, his experience with both the Genexus Purification System and Integrated Sequencer.

In his laboratory, they compared their current “semi-automated workflow using the Ion Chef™ Instrument and Ion GeneStudio™ S5 System for sequencing and semi-automated extraction system, with the new, automated workflow.” They concluded that the new workflow allows them to automate many hands-on steps and streamline the process, shortening their turnaround time (TAT) from 4 to 2–3 days.

In his detailed comparison of the two extraction systems, the Promega Maxwell RSC™ 48 and the Genexus Purification System, Dr. Seidman highlighted that while both require some preprocessing of the sample and take up similar amount of bench space, the Genexus platform also performs the quantitation of the DNA and RNA. While the Promega platform can extract 48 samples, it can only extract DNA or RNA one at a time. The Genexus platform can process up to 12 samples at a time, but can extract both DNA and RNA at the same time, resulting in 24 quantified samples (12 RNA and 12 DNA).

Dr. Seidman also discussed the Genexus Software, highlighting the feature which allows the lab to choose their own quality control (QC) criteria for a successful sample and then easily review if these were met or not.

In conclusion, he highlighted the fast TAT and significant reduction of hands-on time, reducing potential for variability in results.



To view Dr. Seidman's presentation, please visit the Ion World on-demand video at oncomine.com/genexus-oncology

Experience from the world of easy NGS



During a webinar in collaboration with The Pathologist Journal in October 2021, we introduced more new users of the Genexus Integrated Sequencer and Oncomine™ Precision Assay.

Among these were Annarita Destro, PhD, senior molecular biologist at Humanitas Research Hospital in Italy, and Reiltin Werner, MSc, chief medical scientist in the Molecular Pathology Laboratory at Cork University Hospital in Ireland. Both have newly introduced NGS into their laboratories with the Genexus Integrated Sequencer having previously used only single-gene testing methods.

The need to use NGS to consolidate biomarker testing in certain tumor types with limited tissue sample, as the number of biomarkers to be tested grows, was highlighted by both presenters. They also agreed that the Genexus System made it easier to adopt NGS technology than they first thought.

As part of their NGS verification process, Drs Destro and Werner reported high concordance between the Genexus System and other methods, as well as intra and inter-run reproducibility results using different tumor samples (Figure 3).

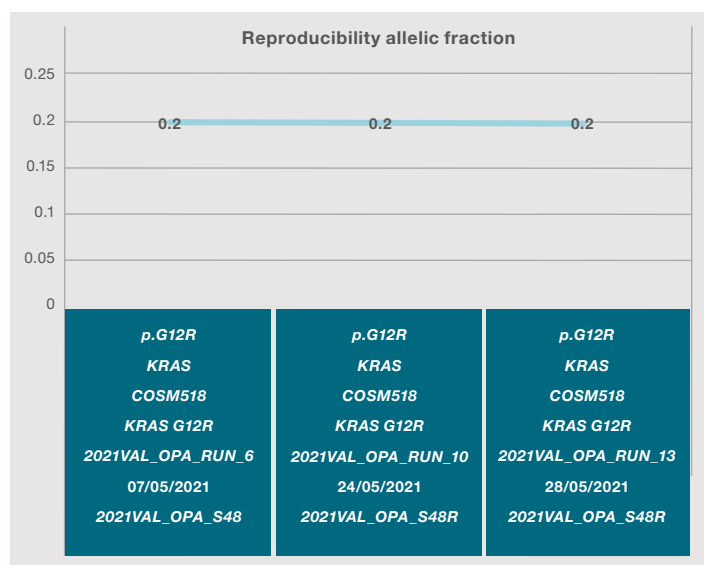


Figure 3: Reproducibility assessment in 3 runs by 3 different operators on 3 different days—results consistent with an AF of 20% for *KRAS G12R* in the Molecular Pathology Laboratory at Cork University Hospital.

Dr. Destro also presented her lab's experience with the Genexus Software from run setup to final results and commented on the ease of use without the need for bioinformatics expertise.

Watch the webinar on demand on oncomine.com/genexus-oncology

Combining oncology biomarker testing with SARS-CoV-2 surveillance in times of crises

Over the last 20 months, the need for SARS-CoV-2 testing has been dominating the laboratory community. To secure the capacity required by the situation, many labs that traditionally have not been involved in virology, but have experience in molecular testing have stepped in and helped out carrying on either PCR or sequencing SARS-CoV-2 tests. Among them is the Laboratory of Clinical and Experimental Pathology at Louis Pasteur Hospital in Nice, France. The head of the laboratory,

Paul Hofman, MD, PhD, presented his experience at Ion World 2021.

While they already use the Oncomine Precision Assay for clinical research testing of key relevant biomarkers, they have embarked on a project to evaluate the Ion AmpliSeq™ SARS-CoV-2 Insight Research Panel for prospective surveillance testing, using the Pangolin software for analysis.

RETROSPECTIVE ANALYSIS

STARTED MARCH 2021

From RT-PCR to NGS
assessment of SARS-CoV-2

145 nasopharyngeal (NSP) swabs samples and 145 matched saliva samples RT-PCR positive for SARS-CoV-2

Individuals tested from April 2020 to January 2021

Samples collected and stored at -80°C in the biobank 0033-00025

PROSPECTIVE ANALYSIS

STARTED APRIL 2021

Systematic surveillance of NSP swabs positive for RT-PCR-SARS-CoV-2 but without any variant identification using the commercial multiplex testing

Testing of NSP swabs with clinically suspected SARS-CoV-2, but negative for RT-PCR-SARS-CoV-2

Testing of NSP swabs in some specified clusters



Summarizing his laboratory experience with the Genexus Integrated Sequencer, Paul Hofman stressed the advantages of its automation requiring less trained personnel to operate it. During the times of crisis, with social distancing requirements, the ease of use of the Genexus System and the ability to provide results in approximately 24 hours, has proven to be beneficial to the lab.

In his own words, such advancements in NGS technology make it possible for smaller, local laboratories to provide genomic profiling services across different applications.

He stated that they are able to process up to 112 samples on the Genexus Integrated Sequencer in a single week using the Oncomine Precision Assay for tumor biomarker testing. The assay covers genes of interest for most solid tumours (notably lung cancer). He also highlighted high sensitivity of variant detection (ability to detect variants even with a low viral load/high Ct) of the Ion AmpliSeq SARS CoV-2 Insight Research Panel.

To view Paul Hofman's presentation, please visit the Ion World on-demand video at oncomine.com/genexus-oncology

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More complete myeloid genomic profiling.

- End-to-end workflow automation, from nucleic acid extraction to annotated variant report*
- Specimen-to-report turnaround time of 1 to 2 days, with only 20 minutes of hands-on time required
- 45 DNA and 30 RNA gene targets relevant for myeloid malignancies
- Additional genes added to the panel to further improve coverage of key targets
- Enhanced performance for challenging-to-sequence targets like *CEBPA*, *FLT3-ITDs*, and more

[Watch the video >>](#)

* The automated end-to-end workflow will be available for RNA samples after the Total RNA Purification kits are available in approximately early 2022. The content provided herein may relate to products or workflows that have not been officially released or fully validated and is subject to change without notice.

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