OmniTier

COMPSTOR NOVOS® PRO

High Throughput, Secondary Analysis On-Premise, Private Cloud Appliance for Short and Long Read Sequencing

Product Brief

For Research Use Only

Primary Analysis Sequencer

Input: Raw detector signals

Output: FASTQ file(s) with sequencing reads and quality scores

Secondary Analysis Variant Identification

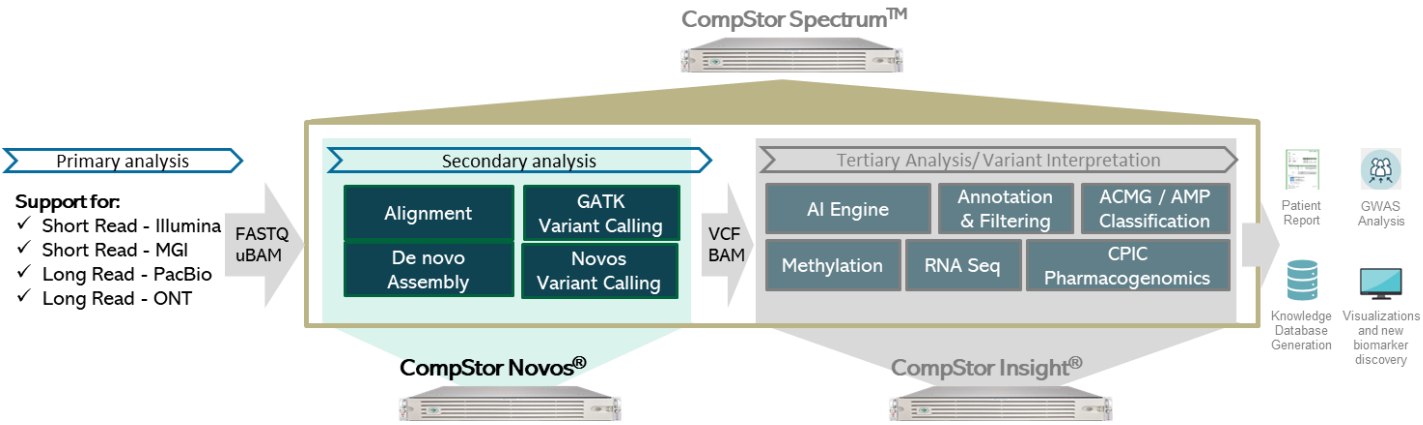
Input: FASTQ files from short or long read sequencers

Outputs: 1) BAM file with aligned reads, 2) VCF file with variant calls

Tertiary Analysis Variant Interpretation

Input: VCF, BAM files

Outputs: Reports of interpretation of variants and results



Overview

CompStor Novos® Pro is an accelerated, human genome secondary analysis, on-premise appliance for WGS, WES and targeted gene panels. Supporting short and long read sequencers.

Fast analysis times using proprietary technology provides low cost per genome. Proprietary, highly accurate variant calling is driven by a domain specific, deep learning algorithm, in addition to standard GATK.

Benefits

- ☑ Lowest cost per genome analysis solution. No additional usage fees per analysis.
- ☑ Easy to set up and easy to use GUI. Stand alone, plug-and-play appliance. No scripting required.
- ☑ Protect your investment. Increase throughput by adding more CompStor Novos Pro nodes. Automatically scales performance.
- ☑ Multiple sequencing technologies (Illumina, BGI, PacBio, and Oxford Nanopore) supported on same platform.
- ☑ Both GATK and proprietary variant calling for NGS reads. SNP, indel, structural variants, CNVs and Mitochondrial variant calling

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Product Brief

Secondary Analysis Highlights

CompStor Novos® Pro provides accelerated, human genome secondary analysis for whole genome sequence (WGS), whole exome sequence (WES) and targeted gene panels. Supports both short NGS and long read sequencers.
Fast analysis times using proprietary technology provides low cost per genome. Proprietary, highly accurate variant calling is driven by a domain specific, deep learning algorithm, in addition to standard GATK.

Automated Pipeline	• Fully integrated pipeline appliance that is quick and easy to set up and run with a simple graphical user interface. Standard programming interfaces support batch and remote operation. • Somatic, Germline • WGS and WES
Fast Run Times	• Reference alignment and variant calling in industry leading times, 1-5 hours depending on configuration
Features	• De novo Assembly reconstruction • Alignment-based reconstruction • GATK & OmniTier Novos proprietary variant callers • Support for Illumina, MGI Tech, Oxford Nanopore and PacBio sequencers
Accurate Variant Calling	• Surpasses standard open-source variant calling performance for SNV/SNPs, short Indels, structural variants, CNVs and mitochondrial variants • Detects variants from de novo-assembly output contigs
High Coverage Genomes	• Sample sequence coverage tested up to 300x and input file sizes up to several terabytes
Flexible Job Scheduler	• Automated job scheduler (up to 10,000 jobs) • Data ingress web application for ingressing of FASTQ files from external file storage
Accelerated Preprocessing	• Demux, bcl2fastq • PacBio CCS processor • Preloaded GRCh37 and GRCh38 references • Custom reference bundles supported
Extensible Platform	• Can be installed on-premise, private Edge or software installed in an HPC • Ability to add new features and performance enhancements • Analytics • Customizations

Table 1: Secondary analysis features

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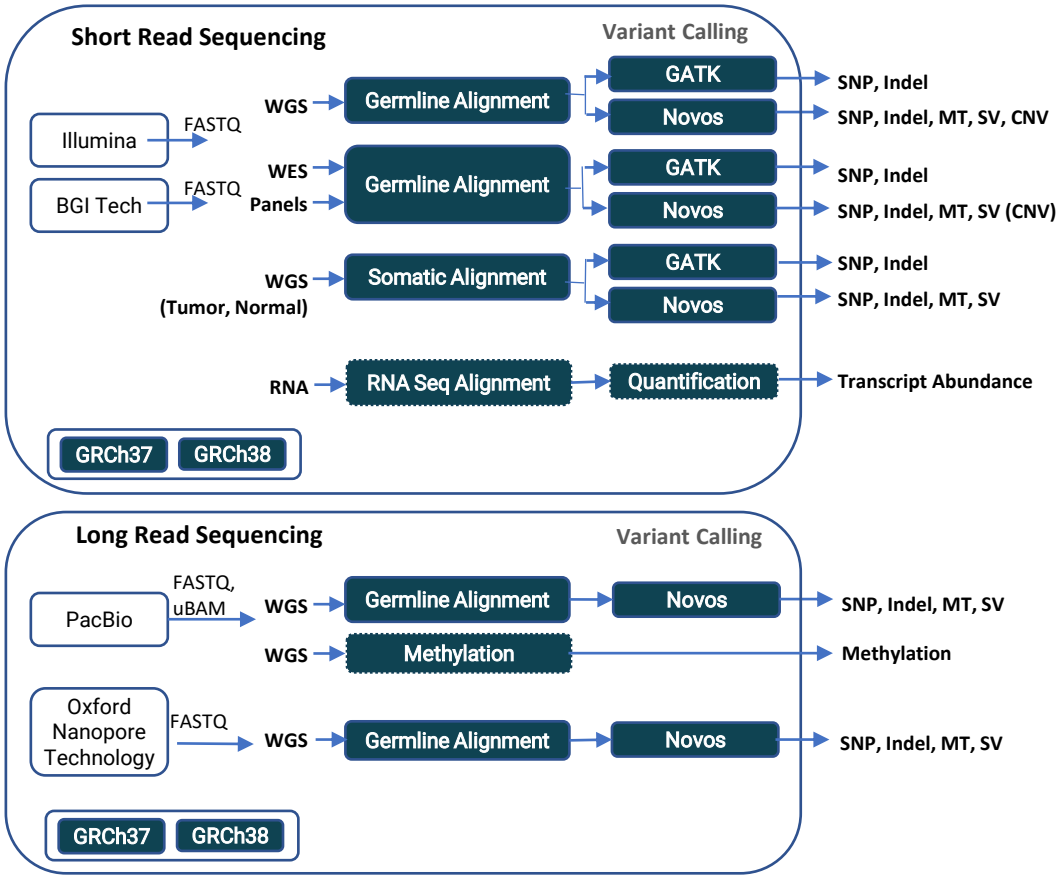
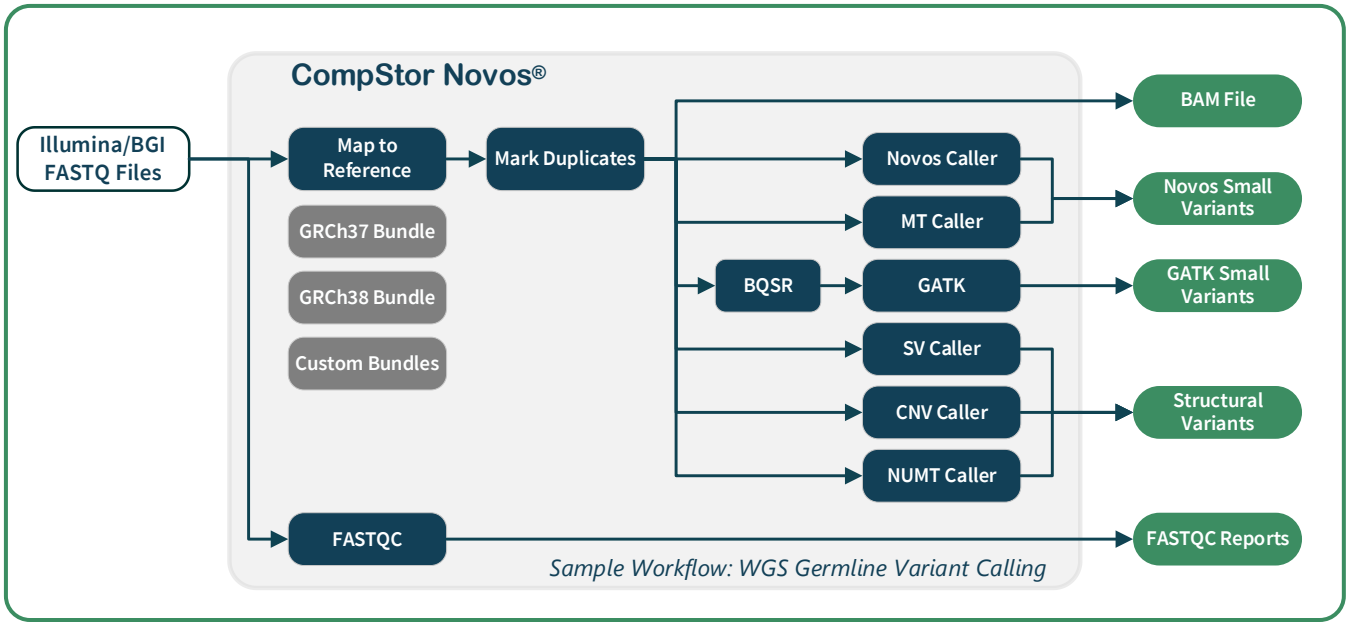


Comprehensive short and structural variants called



Accurate Variant Calling

OmniTier's CompStor Novos® Pro bioinformatics appliance shows greater variant calling accuracy across all seven NIST Genome in a Bottle (GIAB) datasets than GATK and Strelka as measured by F1 score.

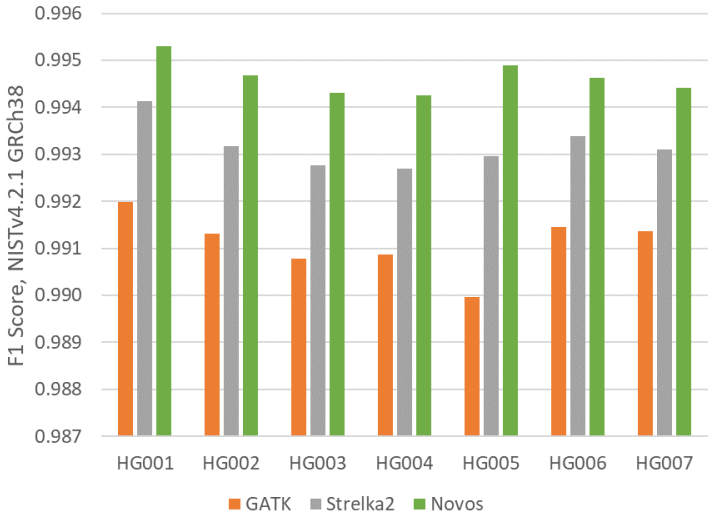


Figure 1: Germline Small Variant Caller Fidelity. WGS reads down-sampled to 35X coverage from publicly available GIAB data. Accuracy is measured against the NIST v4.2.1 high confidence calls using RTG vcfeval.

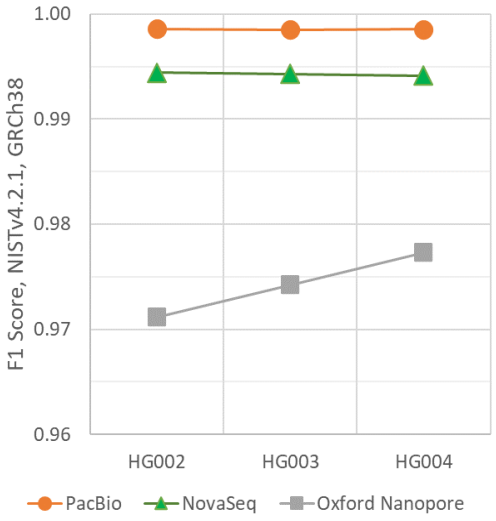
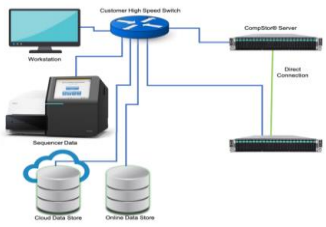
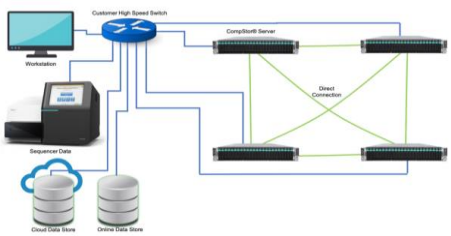


Figure 2: Germline small variant call fidelity using Precision FDA Truth V2 challenge data sets.

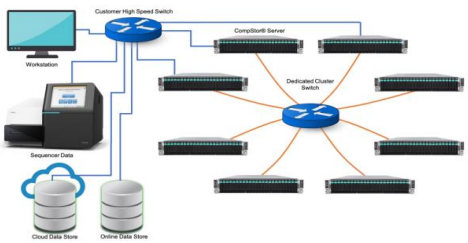
CompStor Novos® Pro Appliance Configuration



2-node configuration



4-node configuration



8-node configuration

On-Premise Appliance, Private Cloud, HPC

For more information please email: sales@omnitier.com

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