OmniTier

COMPSTOR NOVOS®

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

Product Brief

For Research Use Only. Not for use in Diagnostic Procedures.

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

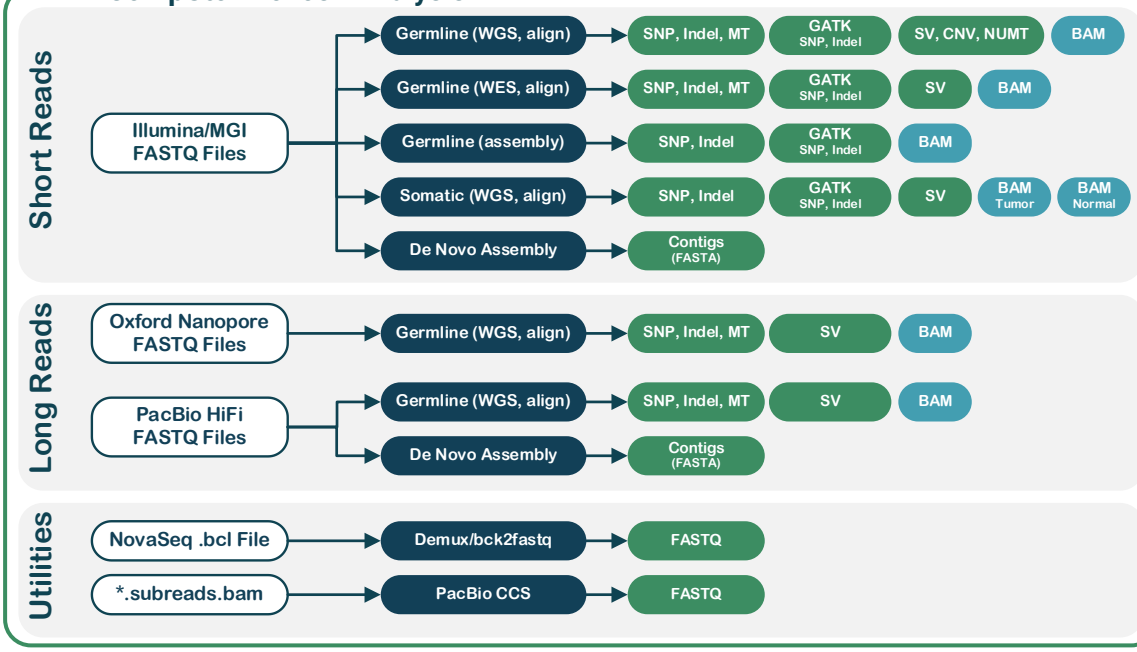
Outputs:1) BAM file with aligned reads, 2) VCF file with variant calls

Tertiary Analysis Variant Interpretation

Input: VCF, BAM files

Outputs:
Reports of interpretation of variants and results

CompStor Novos® Analysis



Overview

CompStor Novos® 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 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

Product Highlights

Automated, Intuitive 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, 1-3 hours depending on configuration

Features

De novo Assembly Reconstruction

Alignment-based reconstruction

GATK & OmniTier proprietary variant caller

Support for Illumina, BGI, ONT and PacBio sequencers

Mitochondrial support

Accurate Variant Calling

Surpass standard open source variant calling performance in SNV/SNPs, short Indels, SVs and CNVs; additionally, detect variants from *de novo*-assembly output contigs.

High Coverage Genomes

Sequence coverage tested up to 300x and file sizes up to several terabytes

Flexible Data Import

Automated job scheduler (up to 10,000 jobs) and data ingress web application for ingressing FASTQ files from external file storage.

Accelerated Preprocessing

Demux / bcl2fastq

PacBio CCS processing

Preloaded GRCh37 and GRCh38 references. Custom reference bundles supported

Extensible Platform

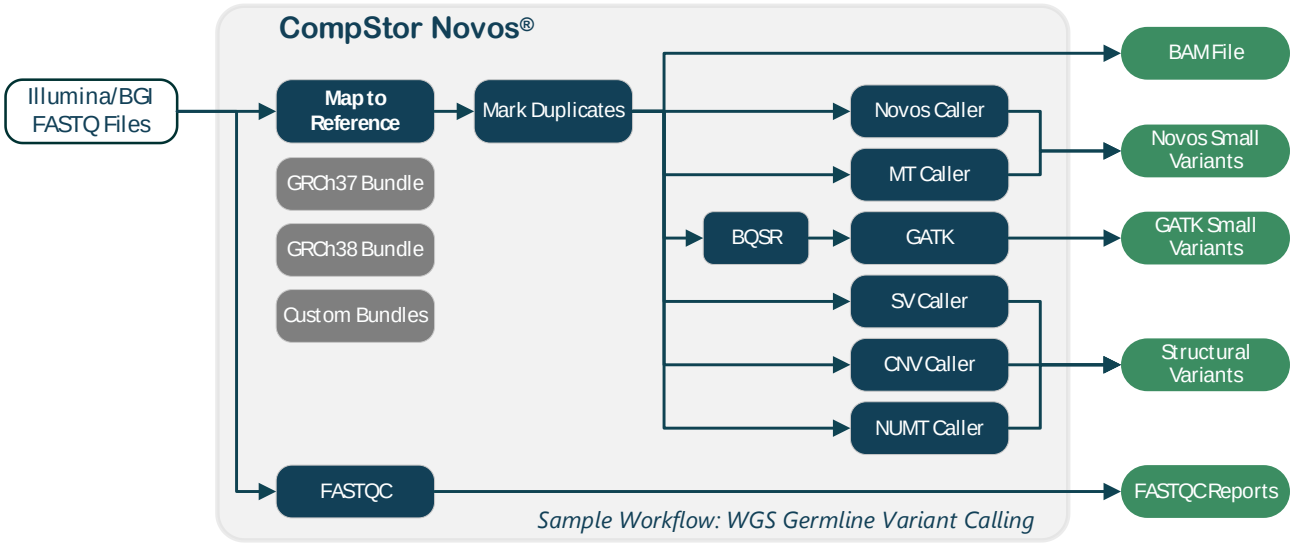
Can be installed on-premise, private Edge, HPC or (in future) in Public cloud

Ability to add new features and performance enhancements

Analytics

Customizations

Comprehensive short and structural variants called



Accurate Variant Calling

OmniTier's CompStor Novos® 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.

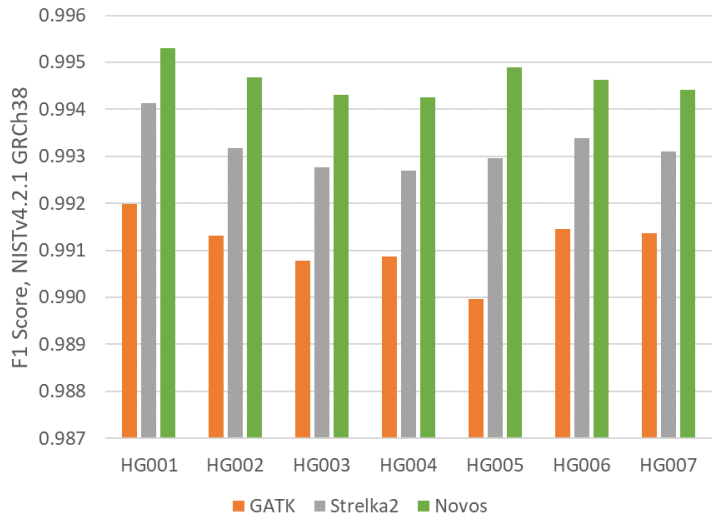


Figure 1: Germline Small Variant Caller Fidelity. WGS reads down-sampled to 35X coverage from publicly available GIAB data. Accuracy is measured against the NIST v4.2.1 high confidence calls using RTG vcfeval.

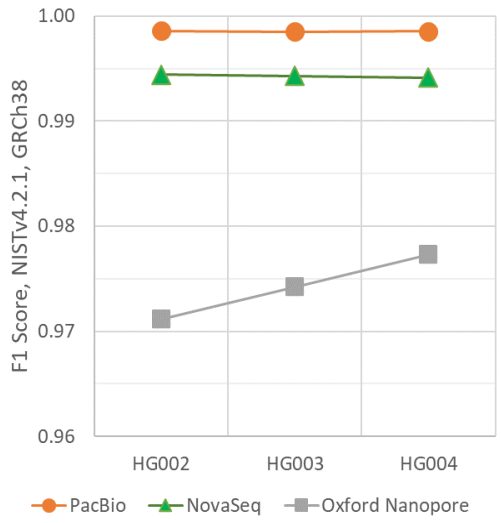
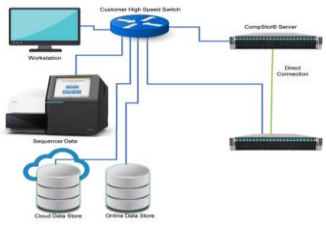
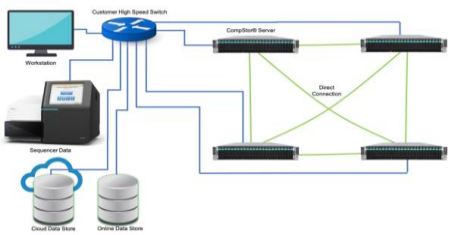


Figure 2: Germline small variant call fidelity using Precision FDA Truth V2 challenge data sets.

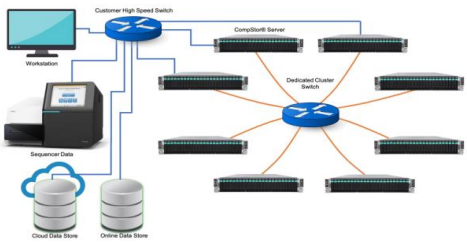
CompStor Novos® Appliance Configuration



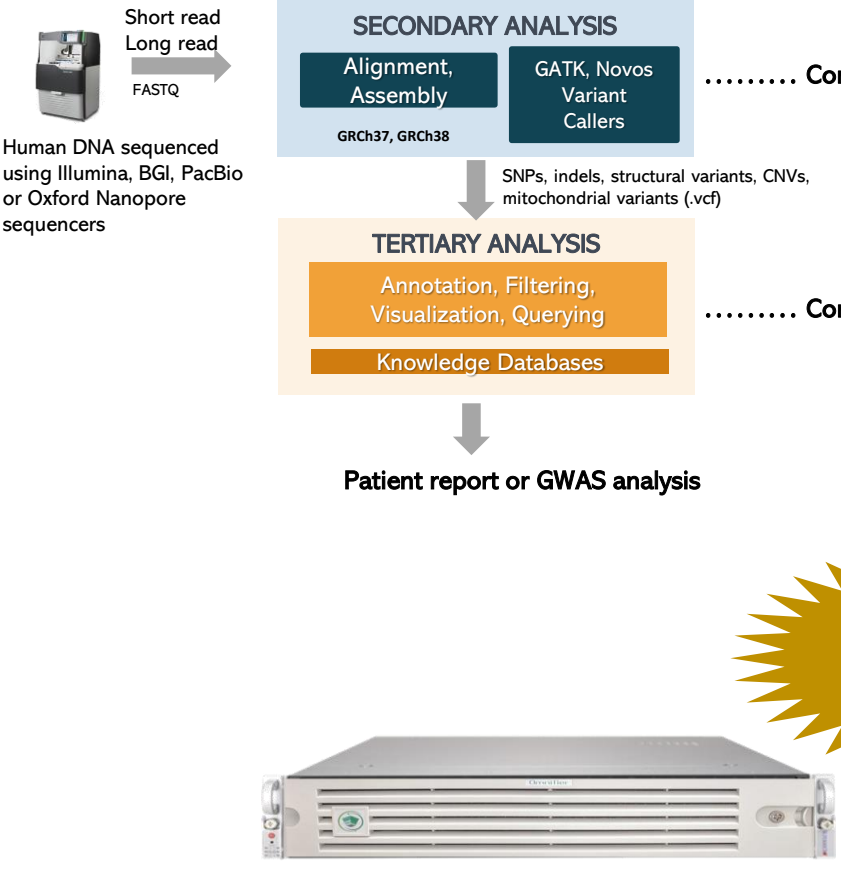
2-node configuration



4-node configuration



8-node configuration



For more information please email: sales@omnitier.com

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