OmniTier

COMPSTOR NOVOS®

High Throughput, Genome Secondary Analysis Appliance

Product Brief

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

High-Throughput Genome Bioinformatics Appliance

Primary Analysis

Input: Raw detector signals

Output: FASTQ file with sequencing reads and quality scores

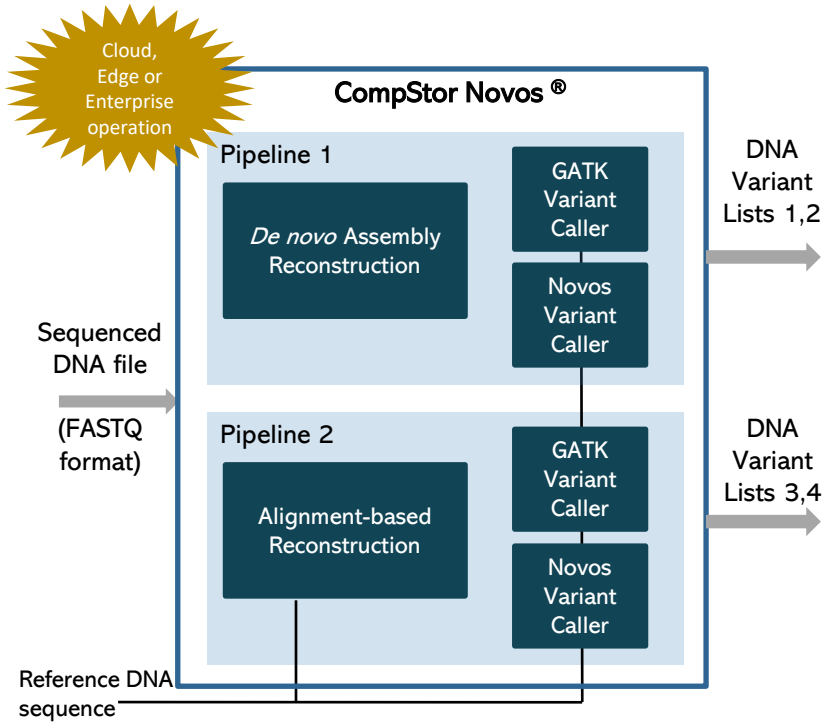
Secondary Analysis

Input: FASTQ files

Output:
i) BAM file with aligned reads
ii) File with Variant calls

Tertiary Analysis

Diagnosis and interpretation of variants and results



POWERING PRECISION MEDICINE

- ✓ **EXPANDED Variant Discovery**
- ✓ **ACCURATE Variant Calling**
- ✓ **HIGH-SPEED Run-Times**
- ✓ **AUTOMATED Pipelines**

Overview

CompStor Novos®. is a Whole Genome Sequencing (WGS) informatics pipeline solution with highest accuracy, lowest latency, and lowest cost per genome bioinformatics appliance with fast turnaround time. Variant calling accuracy exceeds *precisionFDA Challenge* winning results. It is a dual-pipeline enabling *de novo* assembly and reference alignment based methodologies.

CompStor Novos®. is a scalable, multi-node cluster solution that can be installed in an enterprise, Edge or Cloud network and achieves super-computer-class performance in computation times using proprietary algorithms. The cluster configuration can adapt to meet a prescribed range of throughput requirements. An application based job scheduler automates batch processing and allows for a customized analysis workflow.

CompStor Novos®. variant calling is driven by a domain specific, deep learning algorithm that outperforms GATK Best Practices as well as alternative open source pipelines.

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

De novo assembly and variant calling in industry leading times, 1-3 hours depending on configuration

Scalable WGS throughput for de novo processed personal genomes. Standard de novo assembled genomes can realize 3-5X reduction in cost per genome.

Features

De novo Assembly Reconstruction

Alignment-based Reconstruction

GATK & OmniTier Proprietary Variant Caller

OmniTier Proprietary Somatic Variant Caller

Illumina, BGI and PacBio sequencing

Mitochondrial support

Accurate Variant Calling

Surpass standard open source tools in SNV/SNPs, short Indels and CNVs with alignment-based approaches; additionally, detect variants from *de novo*-assembly output contigs.

Extensible Platform

Can be installed in a Cloud, Edge or Enterprise environment

Ability to add new features and performance enhancements

Analytics

Custom services

High Coverage Genomes

Sequence coverage tested up to 300x and file sizes up to several terabytes with 8-node appliance

Flexible Data Import

Automated job scheduler and data ingress web application supporting: FASTQ files from external client or FASTQ files residing on the CompStor Novos® appliance node

Accelerated Preprocessing

Demux / bcl2fastq

PacBio CCS processing

Preloaded and custom reference bundles

Variant Calling Performance Highlights

Accuracy in Variant Calling

OnimTier's CompStor Novos® bioinformatics appliance shows greater variant calling accuracy across all seven NIST Genome in a Bottle (GIAB) datasets than GATK Best Practices pipeline as measured by F1 scores and total errors. Alignment and Assembly results for HG001 are shown in Figure 1. F1 scores are calculated from the fraction of true variants detected (recall) and the fraction of the variants called that are true (precision). In addition, CompStor Novos® F1 scores are higher than all winning entries from the most recent PrecisionFDA Truth Challenge III.

Variant calling utilizes a domain-optimized deep learning methodology to produce fewer false positives and more true positives.

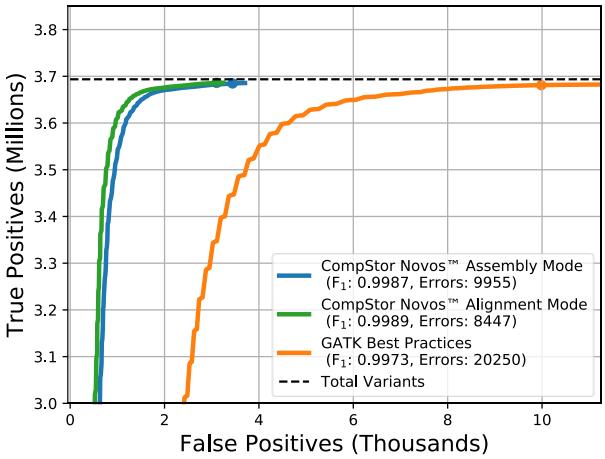
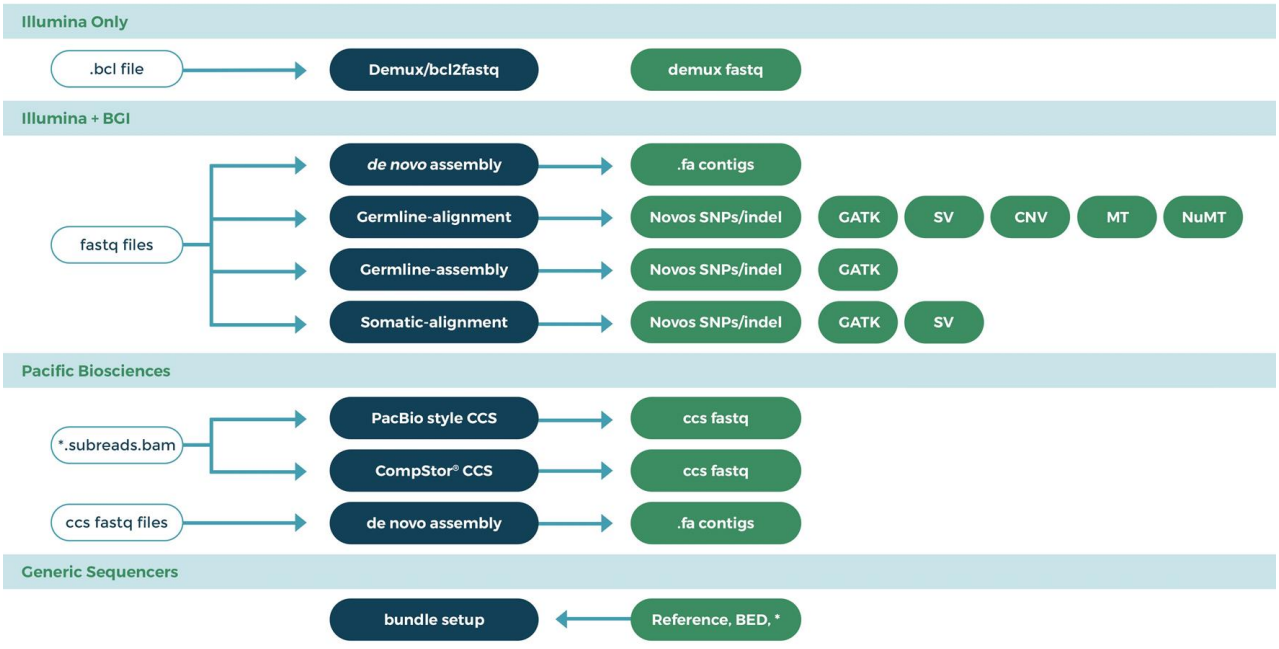


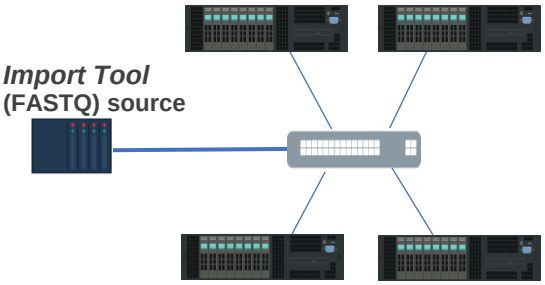
Figure 1 Receiver Operating Characteristics (ROC) for CompStor Novos® versus GATK Best Practices pipeline for HG001 short variants.

CompStor Novos® Feature Set



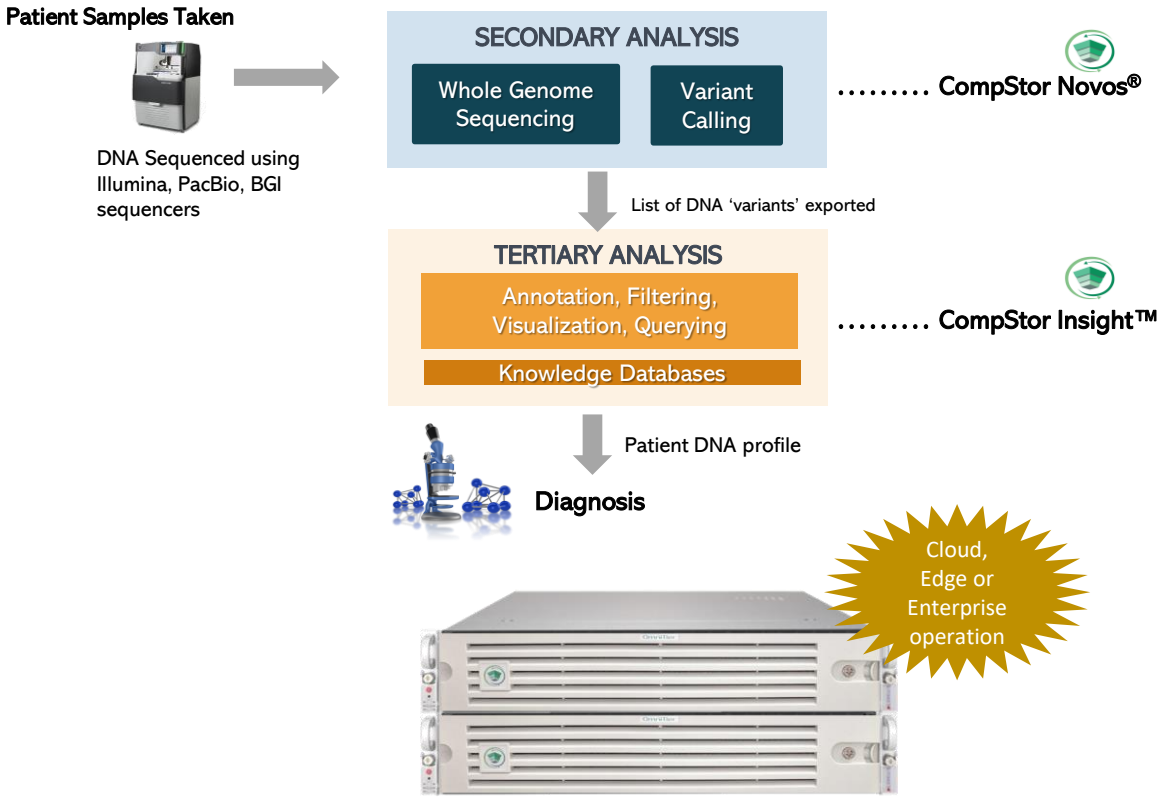
CompStor Novos ® Appliance Configuration

Options (1, 2, 3, 4+ nodes)



Assembly		Alignment	
Configuration	Run-time (hours)	Configuration	Run-time (hours)
2 nodes	3.0	2 nodes	**1.8**
4 nodes	2.0	4 nodes	**1.0**
8 nodes	1.0	8 nodes	**<1.0**

CompStor Novos® scalable, multi-node cluster — High speed data ingress, optimized memory tiers and multi-node communication drive *de novo* assembly and subsequent variant calling. Run-time estimates above are for 35x average coverage depth.



For more information please email: sales@omnitier.com

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