OmniTier

COMPSTOR INSIGHT®

Tertiary Analysis, Variant Interpretation On-Premise Appliance

Product Brief

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

Primary Analysis
Sequencer

Input: Raw detector signals

Output: FASTQ file with sequencing reads and quality scores

Secondary Analysis
Variant Identification

Input: FASTQ files

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

Figure 1. Patient sample to variant interpretation

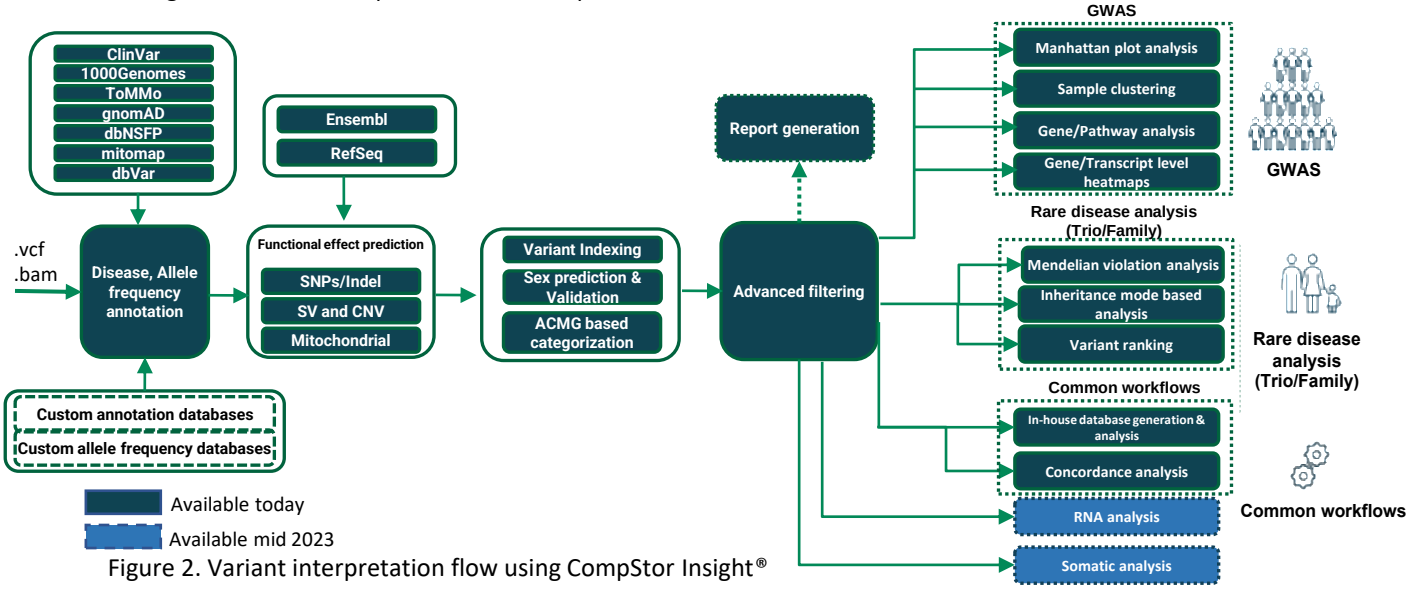

Figure 2. Variant interpretation flow using CompStor Insight®

Overview

CompStor Insight® is an on-premise, high performance, variant interpretation appliance that can also be operated in a private cloud or an HPC. By using OmniTier's proprietary, tiered-memory based indexing and processing, CompStor Insight® accelerates variant interpretation workflows. CompStor Insight® can analyze single or trio gene panels, WES or WGS samples and generate draft reports with prioritized, classified, pathogenic and likely pathogenic variants within minutes.

Benefits

- ☑ Automated and integrated variant interpretation workflow. No scripting! – simpler to get results.
- ☑ Can process 1,000s of samples simultaneously, gaining new insights much faster.
- ☑ Highest data security – all personal data resides on premise, no slow uploading of clinical data to the cloud. No outside internet connection required.
- ☑ Support for custom workflows, knowledge databases, and analysis to integrate seamlessly with existing processes and research areas.

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Product Highlights

CompStor Insight® has been specially developed to accelerate the analysis and diagnostic needs of genomic research centers, academic institutions, government laboratories, hospitals, pharmaceutical, biotechnology, commercial molecular diagnostic laboratories, and consumer genomics companies. CompStor Insight® is highly customizable and supports a wide range of variant knowledge databases (ex: ClinVar, gnomAD) and gene databases (Ensembl, MANE). CompStor Insight® integrates a highly efficient query engine to run user defined queries and also incorporates machine learning based prioritization to identify variants of interest.

Annotation	• Import and annotate against user or custom knowledge databases • Annotates known diseases and allele frequencies based on publicly available datasets • Automated publications, articles and paper lookups in PubMed and Genomenon Mastermind®
Functional effect and pathogenicity prediction	• Predict biological effect based on Ensembl and MANE databases and report RefSeq transcripts when there is match • User modifiable pathogenicity prediction of variants based on ACMG-AMP guidelines[1] • Analysis of variants for ACMG incidental reporting[2]
Filtering	• Support for gene panels, whole genome sequencing (WGS) and whole exome sequencing (WES) • Filter by user defined queries, Quality control • Identify candidates of interest
Single sample analysis	• Sex prediction • SNP, Indel, SV, CNV, and NuMT variant analysis • Draft and final report generation. Standard and custom report templates
Trio/family analysis	• Mendelian violation analysis • Inheritance-mode based analysis, categorization, and UPD detection • SNP, Indel, SV, CNV, and NuMT variant analysis • Draft and final report generation. Standard and custom report templates
GWAS analysis	• Most mutated genes across samples by Gene/Gene Pathways /Disease/biological effects • Summarizes genome-wide association data to spot regions of the genome that cross statistical significance thresholds • Sample clustering
Visualization	• Integrated query and visualization of genomic data • Genome viewer to assess the effects of variants • Concordance analysis
Collaboration Tools	• Organize samples and analyses into ‘cases’ • Assign and review cases or variants • Create and share variant level, case level notes

Table 1: CompStor Insight® features

1. https://pubmed.ncbi.nlm.nih.gov/25741868/
2. https://www.nature.com/articles/gim201373

Variant Analysis and Interpretation

CompStor Insight® allows for complete end-to-end variant interpretation from the annotation and filtering of variants of interest all the way through to automated report draft generation. Optimized data processing harnessing sample phenotype information and ACMG guidelines allows for the identification of primary findings and report generation within minutes. After user review, reports can be exported as PDF files. Complete automation of the interpretation flow is accomplished via a convenient Python API.

Figure 3. Case Management Features

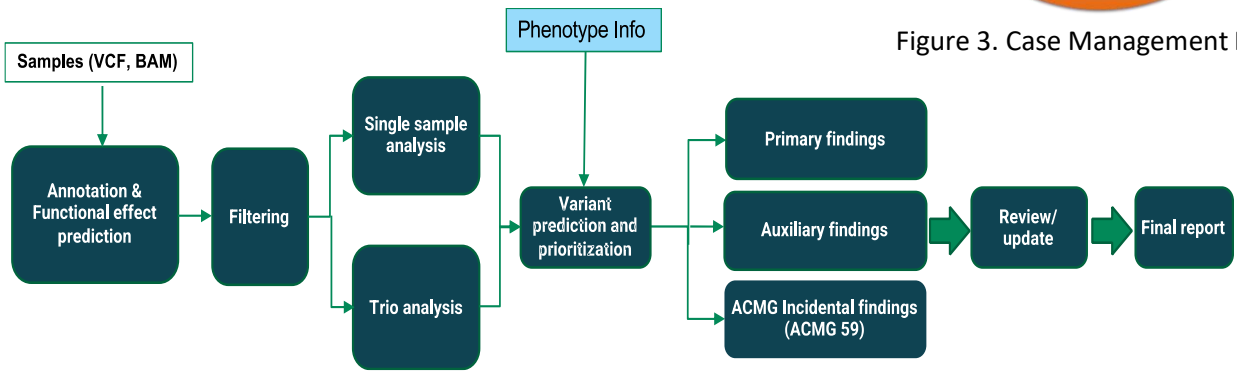

Figure 4. CompStor Insight® report generation workflow

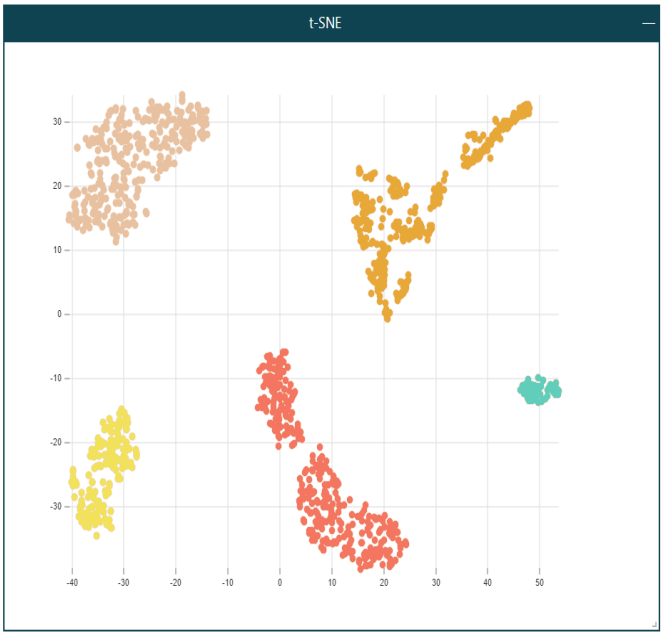

Figure 5. Sample clustering using CompStor Insight®

GWAS Analysis

GWAS analyses including Manhattan Plot generation and clustering analysis allow for efficient large-scale data processing with easy-to-understand results. Additional multi-gene and single-gene statistical tools are also provided.

Clustering analysis includes both SVD and t-SNE flows in addition to custom user-defined grouping of samples for automatic coloring within clustering visualizations. The comprehensive variant filtering capabilities of CompStor Insight® are fully integrated enabling GWAS studies targeting individual genes, diseases, pathways, or functional effects.

Manhattan Plot analysis allows for the discovery of variants statistically significant to a test group relative to a control group speeding up variant discovery.

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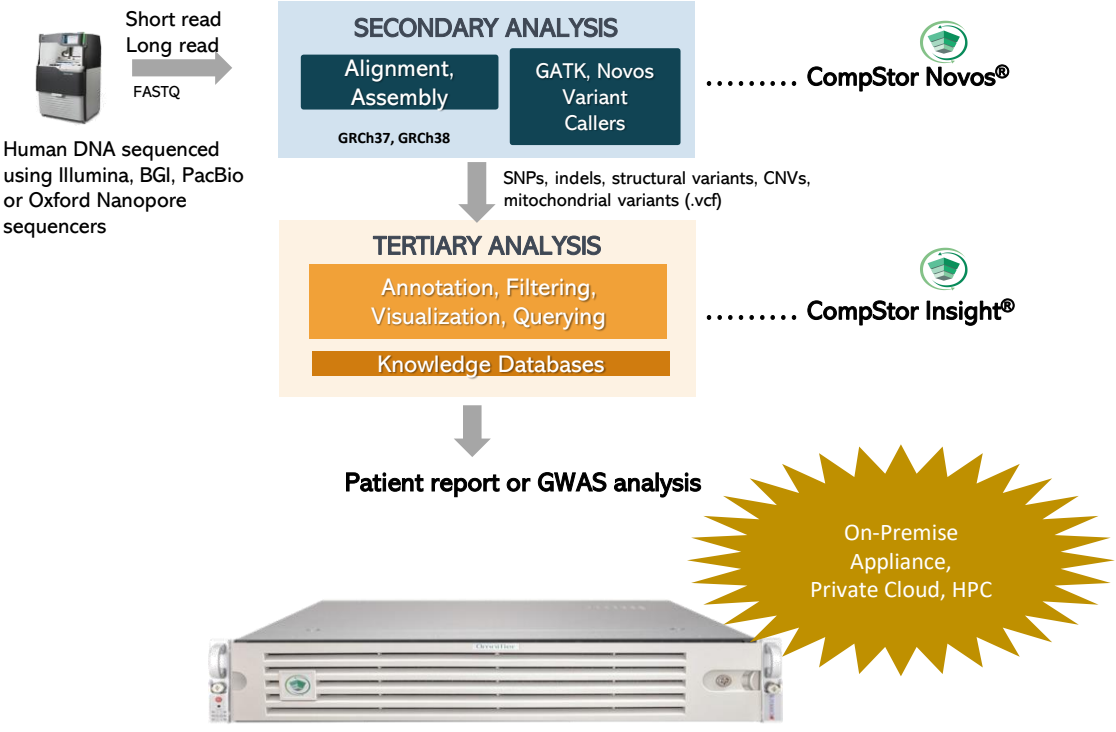
Capacity & Processing Times

CompStor Insight® is a fully optimized end-to-end variant interpretation appliance. A single node of CompStor Insight® can process a trio of WES VCFs and generate a draft report in less than a minute. It comes in a standard 1U form factor and can be connected to customer networks at 1-10 GbE speeds to import patient data securely. CompStor® appliances also provide web-based admin interfaces to manage user accounts, monitor the health of each appliance and to perform software updates.

	WGS	WES
GWAS: Maximum number of samples per analysis	2,000 samples	20,000 samples
Rare Disease Diagnosis: Processing time from .vcf input to draft patient report	7 minutes	< 1 minute

Table 2. CompStor Insight® analysis capacity and processing times



For more information please email: sales@omnitier.com

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