

COMPSTOR INSIGHT®

Tertiary Analysis, Variant Interpretation On-Premise Appliance

Product Brief

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

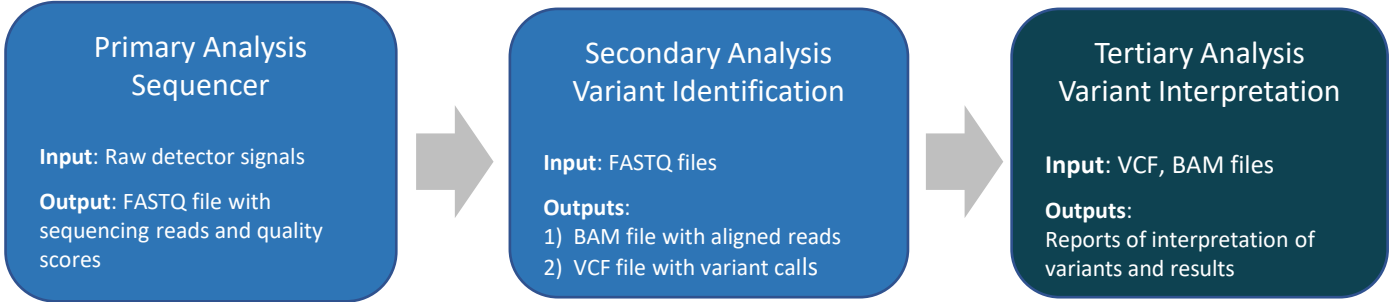


Figure 1. Patient sample to variant interpretation

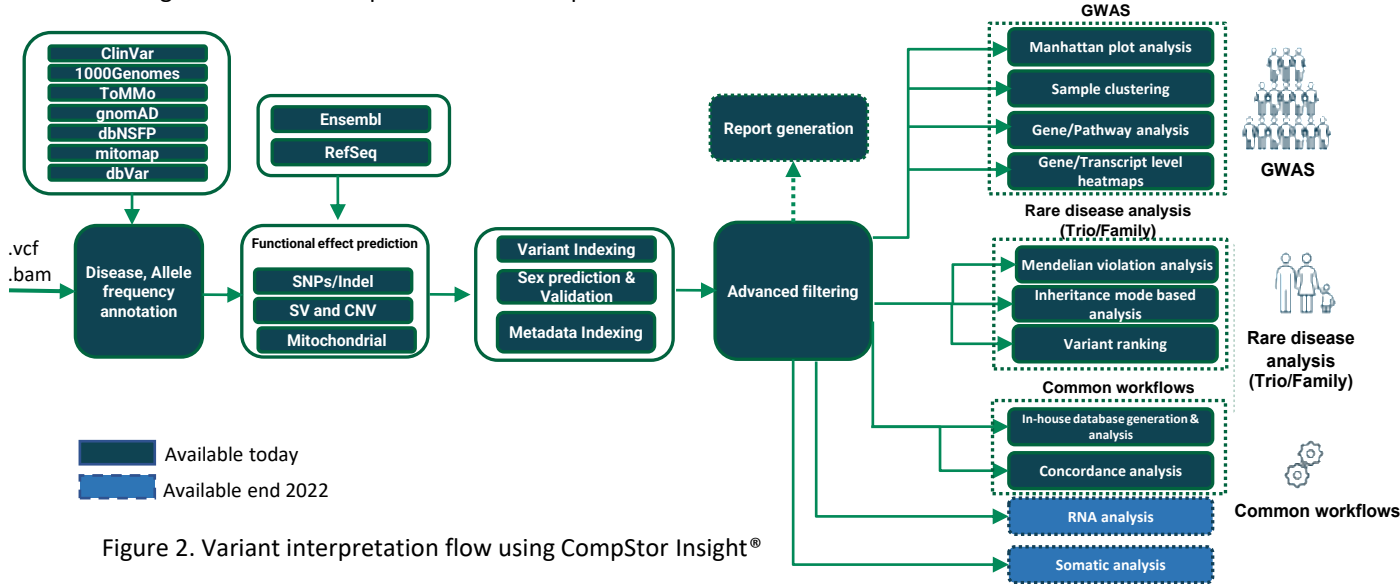


Figure 2. Variant interpretation flow using CompStor Insight®

Overview

CompStor Insight® is an on-premise, high performance, variant interpretation appliance that can also be operated in the cloud or edge. Researchers can use CompStor Insight® to run their variant analysis workflows and interpret the results using an intuitive user interface. By using tiered-memory based indexing and processing, CompStor Insight® accelerates variant interpretation workflows. CompStor Insight® can process a trio of samples and generate draft reports with candidate 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.
- ✓ Support for custom workflows, knowledge databases, and analysis to integrate seamlessly with existing processes and research areas.

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	<ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • 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 guidelines¹ • Analysis of variants for ACMG incidental reporting²
Filtering	<ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • Sex prediction • SNP, Indel, SV, CNV, and NuMT variant analysis • Draft and final report generation. Standard and custom report templates
Trio/family analysis	<ul style="list-style-type: none"> • Mendelian violation analysis • Inheritance-mode based analysis and categorization • SNP, Indel, SV, CNV, and NuMT variant analysis • Draft and final report generation. Standard and custom report templates
GWAS analysis	<ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • Integrated query and visualization of genomic data • Genome viewer to assess the effects of variants • Concordance analysis
Collaboration Tools	<ul style="list-style-type: none"> • 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>

Product Brief

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

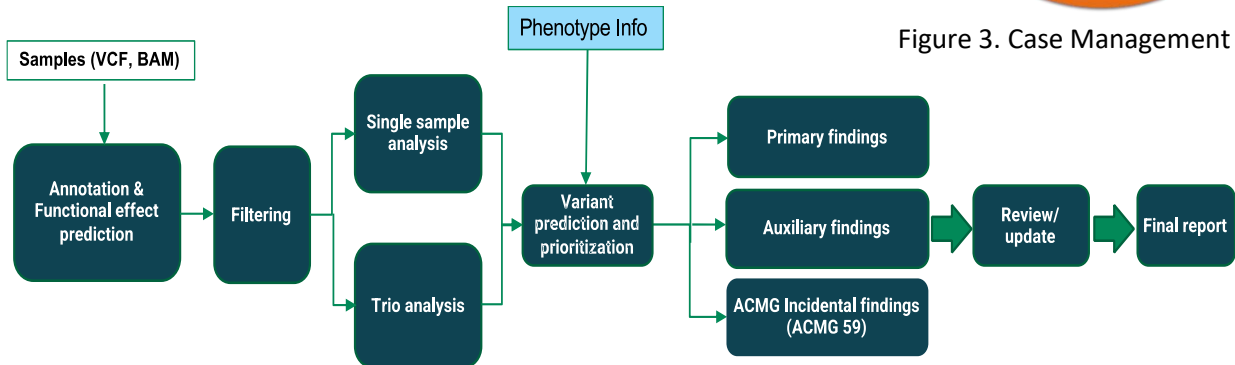


Figure 4. CompStor Insight® report generation workflow

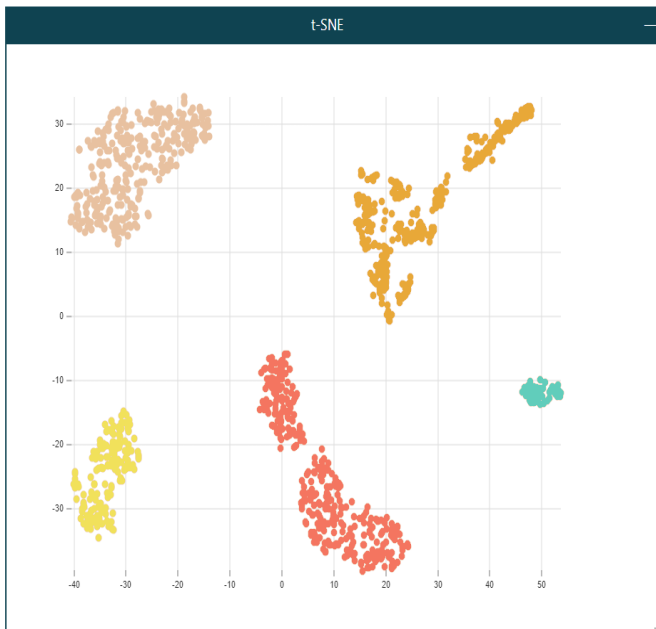


Figure 5. Sample clustering using CompStor Insight®

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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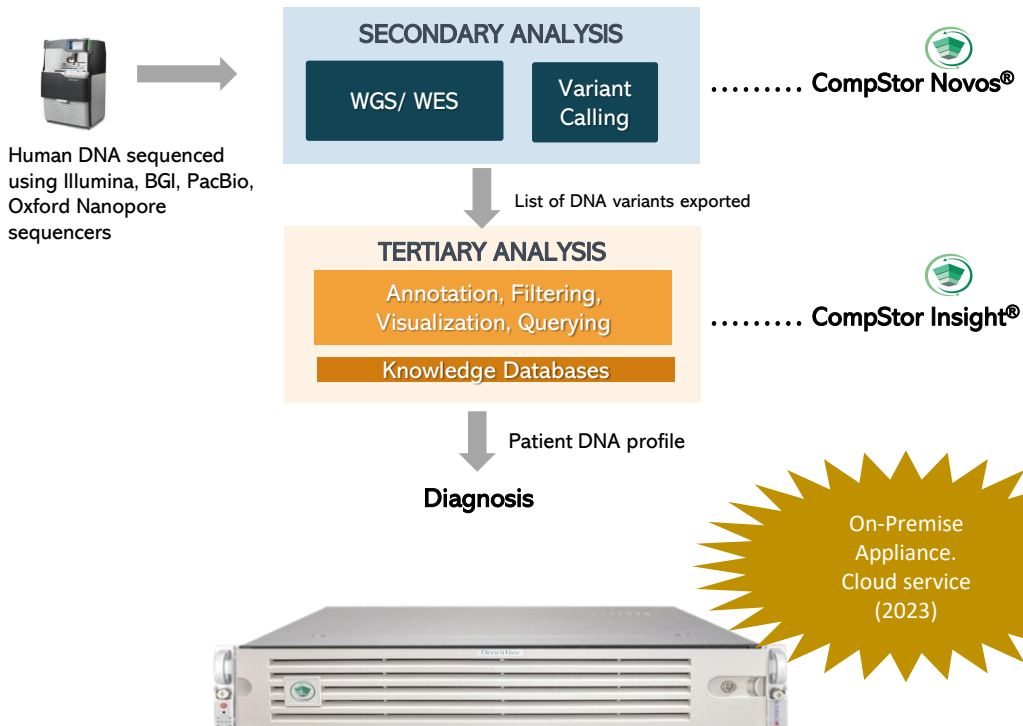
Hardware Platform

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 2U form factor and can be connected to customer networks at 1-10 GB/s 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 SW updates.

	Single Analysis Capacity	Processing Time
WGS	2,000 samples	7 minutes
WES	20,000 samples	< 1 minute

Table 2. CompStor Insight® analysis capacity and processing times (from .vcf input to draft report generation)



For more information please email: sales@omnitier.com

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