OmniTier

COMPSTOR INSIGHT™

Genome Tertiary Analysis Appliance

Product Brief

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

Primary Analysis

Input: Raw detector signals

Output: FASTQ file with sequencing reads and quality scores

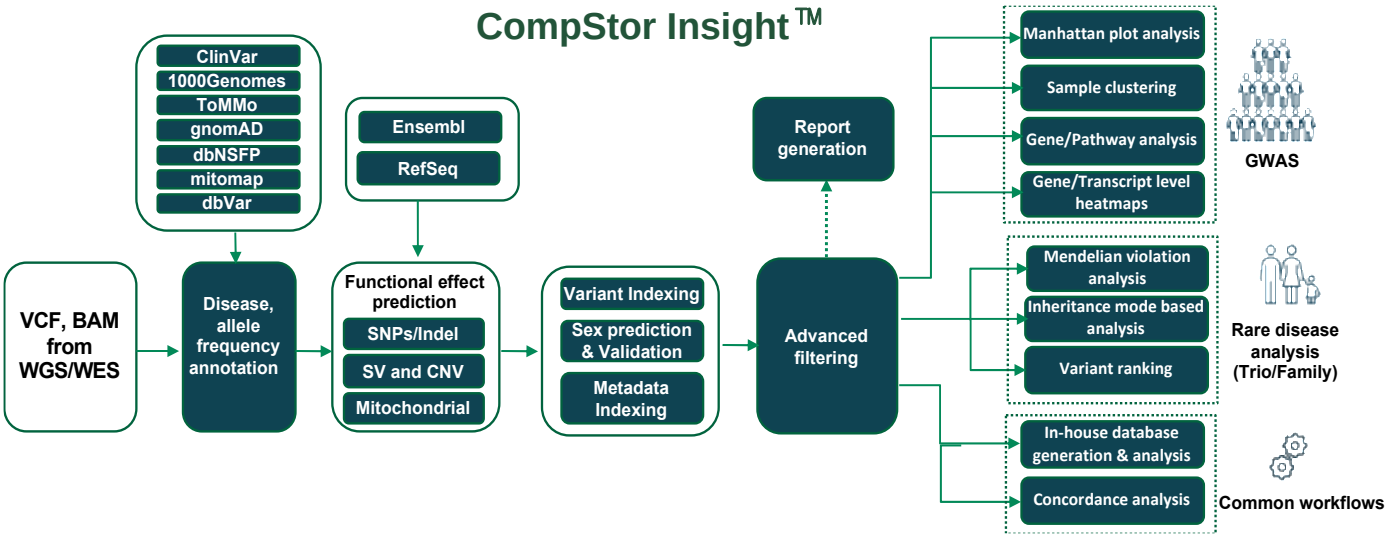
Secondary Analysis

Input: FASTQ files

Output:
1) BAM file with aligned reads and
2) VCF file with Variant calls

Tertiary Analysis

Diagnosis and interpretation of variants and results



Overview

CompStor Insight™ is a high performance, tertiary analysis appliance that can be operated in the Cloud, Edge or on-premises. Researchers can use CompStor Insight™ to run their NGS tertiary analysis workflows and interpret the results using an intuitive user interface. CompStor Insight™ is highly customizable and supports a wide range of variant knowledge databases (ex: ClinVar, gnomAD) and gene databases (Ensembl, RefSeq). CompStor Insight™ integrates a highly efficient query engine to run user defined queries and also incorporates ML based scoring to identify candidates of interest.

Benefits

- ☑ Automated and integrated tertiary 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.

Annotation

Pathogenic status, allele frequencies, variant effect prediction

Seamlessly annotates known diseases and biological effects

Supports germline, SV, CNV, and NuMT VCFs

Identification

Identification of Significant Variants

Manhattan Plot Analysis: Summarizes genome-wide association data to spot regions of the genome that cross statistical significance thresholds

Multi-sample analysis

By Gene: Most mutated genes across samples

By Gene Pathways: Most mutated gene pathways across samples

By Disease: Diseases with most known mutations across samples

By Biological Effects: Most mutated genes, gene pathways with high, moderate or low impact

Trio Analysis : Analysis of de novo mutations for trio samples, Biomarker discovery, Clustering, and Statistical analysis

Flexible Storage

Works with on-premise or cloud storage backends

Automatically tiers data and keeps active data closer to compute

Brings data closer to analysis located at the “edge”

Incorporates advanced caching and data classification technologies to deliver performance

Visualization

Integrated query and visualization of genomic data

Advanced variant query, genome viewer to assess the effects of variants

Zoom into Single Selected Gene

Concordance analysis

Big Data

Variant knowledge databases ClinVar, 1000 Genomes, gnomAD etc.

Gene databases Ensembl, RefSeq, etc.

Can work with custom variant/disease databases

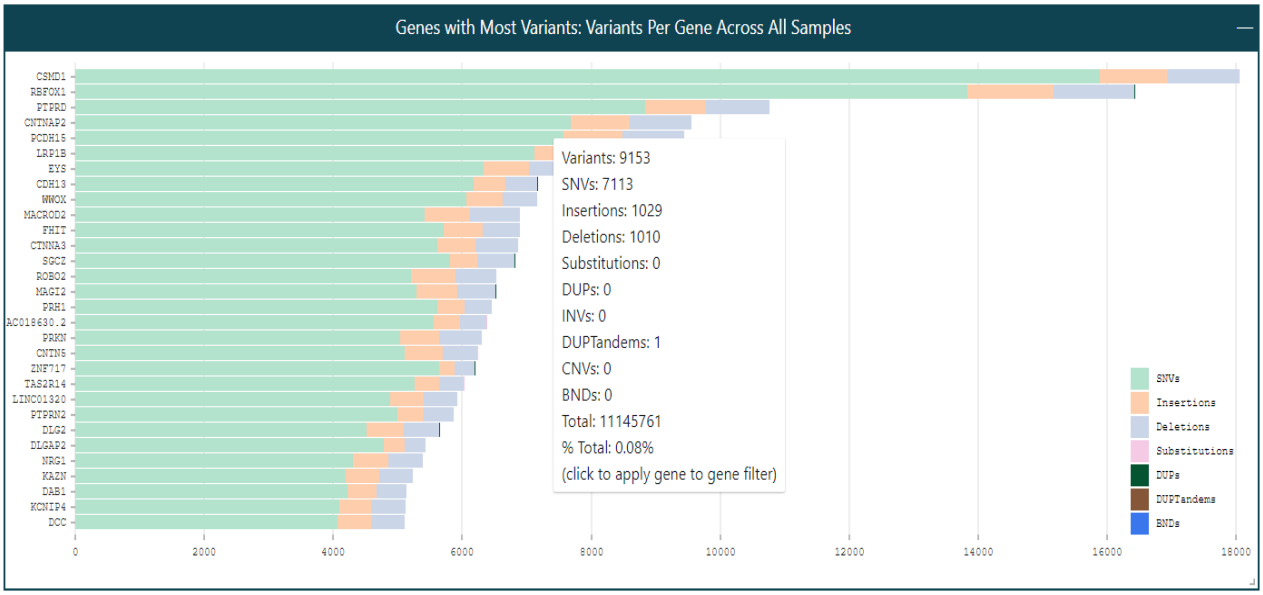
Filtering

Identify candidates of interest

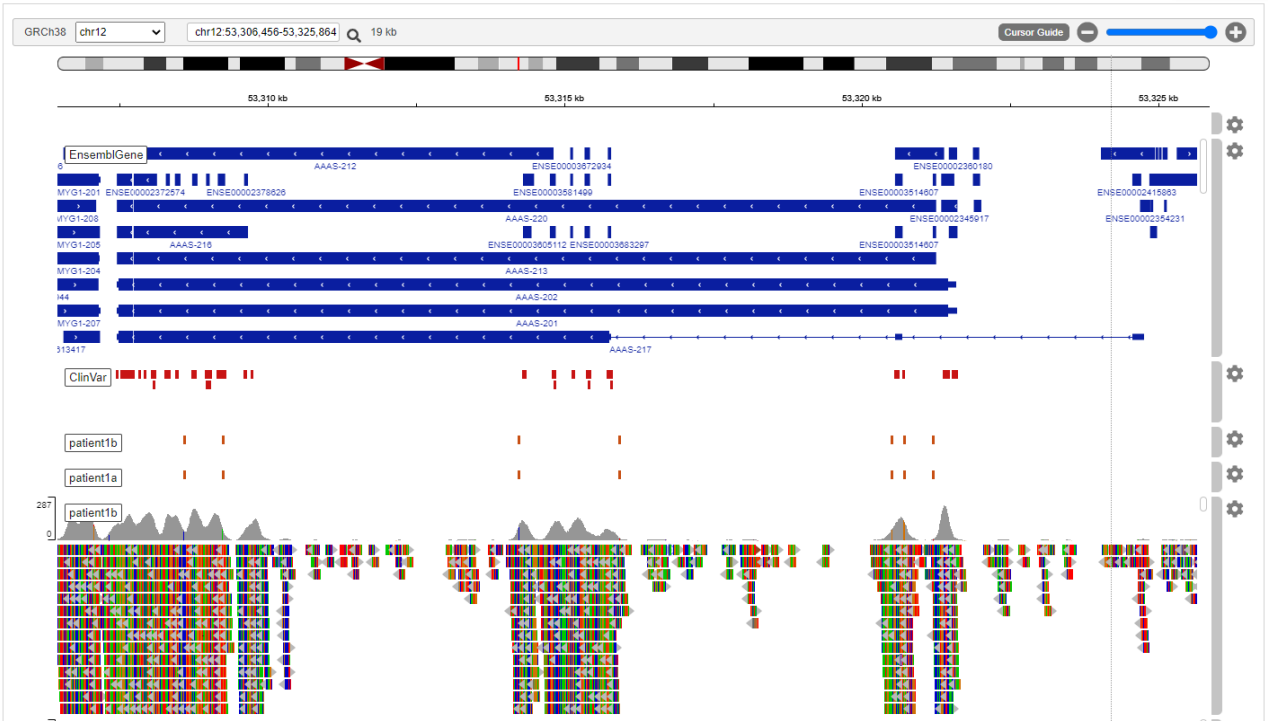
Filter by user defined queries, Quality control

Analytics Highlights

Most Mutated Genes



Genomics Viewer



CompStor® Scientific Computing

Patient Samples Taken

DNA sequenced using Illumina, PacBio or BGI. sequencers

SECONDARY ANALYSIS

Whole Genome Sequencing

Variant Calling

CompStor Novos®

List of DNA variants exported

TERTIARY ANALYSIS

Annotation, Filtering, Visualization, Querying

Knowledge Databases

CompStor Insight™

Diagnostics & Therapy

Automated Knowledge Database Creation

Large GWAS Studies

Patient Profile Creation

Cloud, Edge or on-premise operation

For more information please email: sales@omnitier.com

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