OmniTier

Product Brief

INSIGHT HEREDITARY

Tertiary Analysis essential software for the germline analysis of hereditary and rare disease variants

For use only in CompStor Server

For research use only

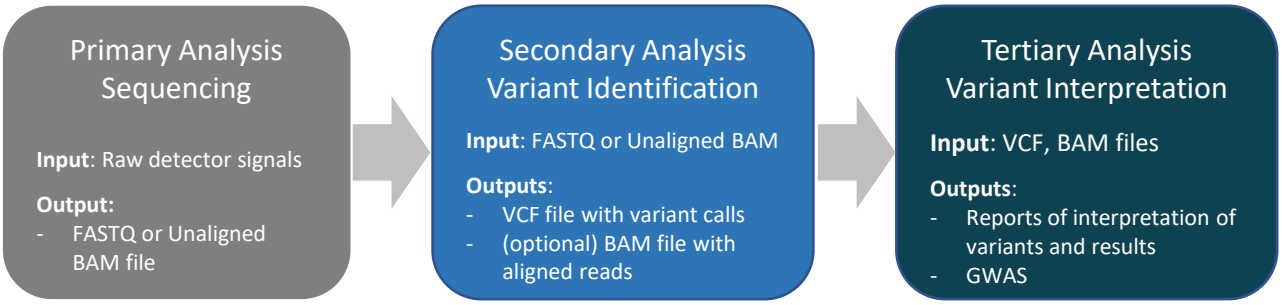

Figure 1: Analysis flow

Overview

Insight Hereditary is germline, hereditary and rare disease tertiary analysis software that runs on OmniTier's CompStor Server platform, either on-premise, in an HPC or in a private cloud. Insight Hereditary can analyze single, duo, trio or larger family collections of gene panels, WES or WGS .vcfs. Outputs can either be draft or final causal reports of prioritized, classified, pathogenic and likely pathogenic variants, or cohorts of samples can be analyzed using GWAS. By using OmniTier's proprietary, tiered-memory based indexing and processing, Insight Hereditary accelerates variant interpretation workflows to provide outputs in just a few minutes.

Variant Analysis and Interpretation

Insight Hereditary 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 and automated prioritization of primary findings and report generation within minutes. After user variant curation, reports can be exported as PDF files or XML. Complete automation of the interpretation flow is accomplished via an available Python API.

Benefits

- ☑ Higher accuracy identification of causal variants
- ☑ Automated and integrated variant interpretation workflow. No scripting! – simpler to get results.
- ☑ Highest data security – No external internet connection required. 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, research and infrastructure.

International Organization for Standardization ISO 13485 Quality Management System

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Insight Hereditary Tertiary Analysis Features and Functionality

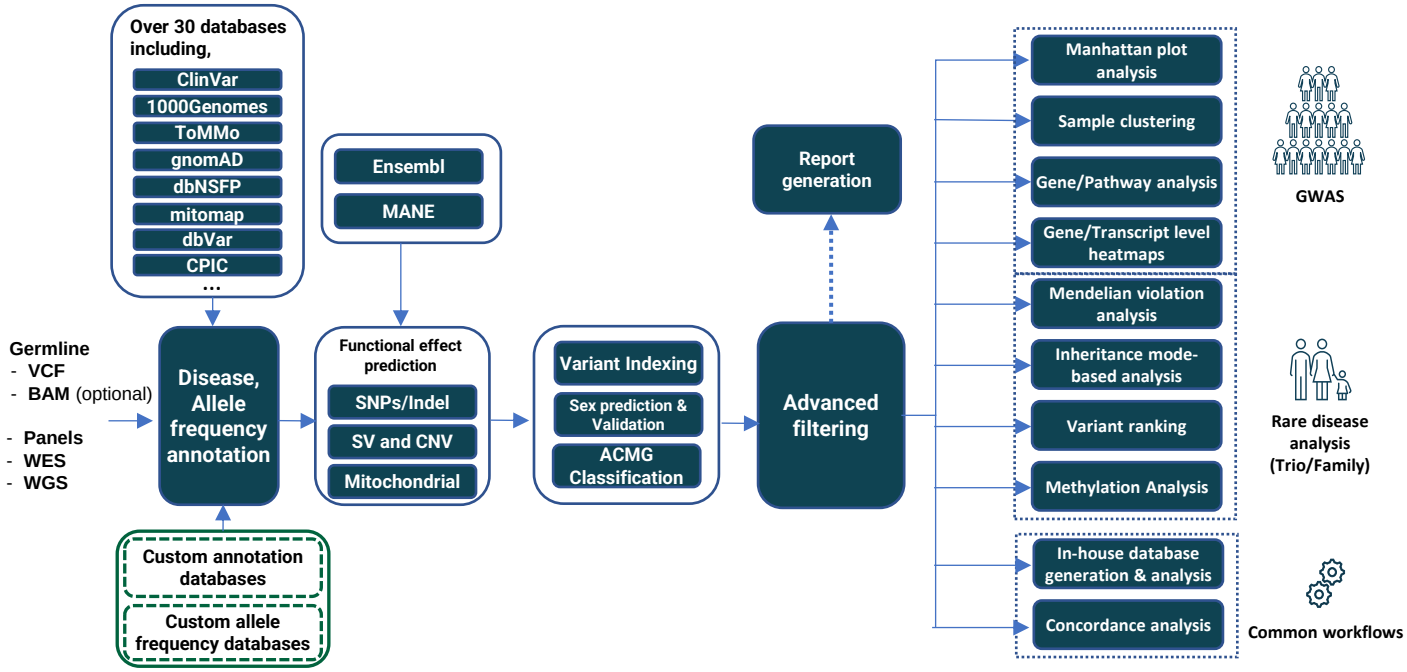

Figure 2. Insight Hereditary variant interpretation flow

Details

	Insight Hereditary (Pro)	Insight Hereditary (Workgroup)	Insight Hereditary (Personal)
Max number of user accounts	500	50	5
Max number of user-defined workspaces	100	10	1
Germline Sample Analysis	Single, Duo, Trio, Family (>3 samples)		Single, Duo, Trio
Variant report output format	PDF, XML	PDF, XML	PDF only
Python API integration and automation support	Yes	Yes	No

CompStor Server	**Tertiary Analysis Turnaround Time** (.vcf to draft report)	
	WES (100x)	WGS (30x)
CompStor Server Pro	<< 1 minutes	7 minutes
CompStor Server Workgroup	2 minutes	12 minutes
CompStor Server Personal	5 minutes	30 minutes

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Product Highlights

Applications

Germline Rare and Hereditary Diseases	• ACMG Classification
	• Analysis types:
	  ➢ Single, Duo, Trio, Family
	  ➢ Inheritance mode based
	• Uni Parental Disomy (UPD), Loss of Heterozygosity (LOH) detection
	• Mendelian violation detection

Gene Expression	• Methylation analysis
Genome Wide Associate Studies (GWAS)	• Most mutated genes across samples by Gene/Gene Pathways /Disease /biological effects • Genome-wide association data to spot genome regions that cross significance thresholds • Sample clustering
Consumer Genomics	• Polygenic risk scoring

Features

Annotation	• Support for germline gene panels, whole exome sequencing (WES) and whole genome sequencing (WGS) • Annotates .vcfs with known diseases and allele frequencies using over 30 internal knowledge databases along with user-imported databases
Visualization	• Integrated query and visualization of genomic data • Genome viewer to assess the effects of variants • Concordance analysis
Variant Interpretation	• Reference publications lookups in PubMed and Genomenon Mastermind® with automated link outs • Artificial intelligence (AI) based variant prioritization and ranking, splicing site prediction • Natural Language Processing phenotype look-up and matching • Customized, user defined, table-based variant filtering and sorting
Management and Collaboration Tools	• Organize samples and analyses into ‘cases’ • Assign and review cases or variants • Create and share variant level, case level, user and reviewer notes
Speed and Security	• Fast results in just minutes using OmniTier’s proprietary, MACS acceleration technology • All data resides on-premise. No internet connection required.

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Platform Specifications

	CompStor Server Pro	CompStor Server Workgroup	CompStor Server Personal
Form Factor	Rack Server	Rack Server	Desktop Tower PC
Max number of WGS VCFs stored on the platform	20,000 WES or 2,000 WGS	10,000 WES or 1,000 WGS	3,500 WES or 350 WGS
Max number of WGS sample VCFs in a single GWAS analysis	20,000 WES or 2,000 WGS	10,000 WES or 1,000 WGS	3,500 WES or 350 WGS
Recommended maximum number of simultaneous users	10	5	1

OmniTier's Analysis Product Portfolio

CompStor Server Platforms

CompStor Server Pro

CompStor Server Workgroup

CompStor Server Personal

Essential Software
(Pro, Workgroup, Personal)

Novos *Secondary Analysis*	- Whole Exome, Whole Genome - NGS and Long Read Sequencers - SNP, indel, SV, CNV, Mitochondrial
Insight Hereditary *Tertiary Analysis*	- *Hereditary and Rare Disease, GWAS* - *Germline Panels, WES, WGS* - *Single, Duo, Trio, Family*
Insight Oncology *Tertiary Analysis*	- *Tumor only, Tumor with matched normal samples* - *Biomarker discovery*

For more information please email: sales@omnitier.com

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