



VSWarehouse; a scalable, rapid genomic repository solution

Darby Kammeraad
Field Application Scientist Manager



20 Most Promising Biotech
Technology Providers



Hype Cycle for Life sciences

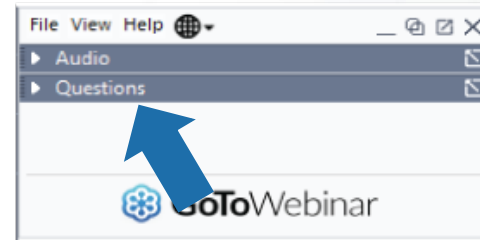


Top 10 Analytics
Solution Providers

Questions & Answers



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- Montana SMIR/STTR Matching Funds Program Grant Agreement Number 19-51-RCSBIR-005
- PI is Dr. Andreas Scherer, CEO Golden Helix.
- The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

Who Are We?

Golden Helix is a global bioinformatics company founded in 1998



Filtering and Annotation

ACMG Guidelines

Clinical Reports

CNV Analysis

Pipeline: Run Workflows



Variant Warehouse

Centralized Annotations

Hosted Reports

Sharing and Integration



CNV Analysis

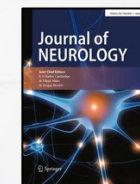
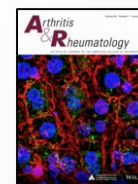
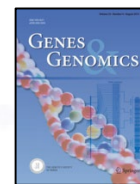
GWAS | Genomic Prediction

Large-N Population Studies

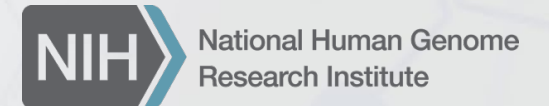
RNA-Seq

Large-N CNV-Analysis

Cited in 1,000s of Peer-Reviewed Publications



Over 400 Customers Globally



When you choose Golden Helix, you receive more than just the software



SOFTWARE IS VETTED

- 20,000+ users at 400+ organizations
- Quality & feedback



DEEPLY ENGRAINED IN SCIENTIFIC COMMUNITY

- Give back to the community
- Contribute content and support



SIMPLE, SUBSCRIPTION-BASED BUSINESS MODEL

- Yearly fee
- Unlimited training & support



INNOVATIVE SOFTWARE SOLUTIONS









- Cited in 1,000s of publications

Gene Panel

Exome

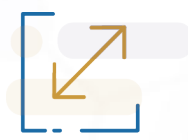
Genome

Sequencer

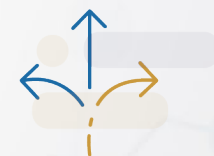
Products	Bioinformatics Pipeline	Function
 DNaseq (Sentieon)  TNseq (Sentieon)  VS-CNV	FASTQ BAM VCF	<ul style="list-style-type: none"> ▶ Single nucleotide variation ▶ Copy number variation & loss of heterozygosity ▶ Chromosomal aberration
Annotations	Annotated VCF	<ul style="list-style-type: none"> ▶ Public & commercial annotations to enrich genomic data sets
 VarSeq  VSReports  VSPipeline	Clinical Report	<ul style="list-style-type: none"> ▶ Annotate & filter ▶ Visually inspect alignments ▶ Variant prioritization ▶ Clinical assessment
 VSClinical	Automated ACMG Guidelines	<ul style="list-style-type: none"> ▶ Clinical variant interpretation in concordance with ACMG Guidelines
 VSWarehouse	Data Warehousing Web-Enabled Interface + Powerful API: JSON, XML, TSV, CSV, SQL, FHIR	<ul style="list-style-type: none"> ▶ Clinical assessment catalog ▶ Advanced data querying ▶ Versioning ▶ Interoperability ▶ Compliance with HIPPA, CLIA & CAP data discovery



Simple



Flexible



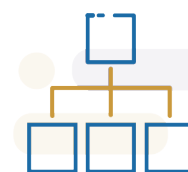
Scalable



Variant annotation
filtering, and interpretation



Powerful GUI with
rich visualizations



Repeatable
workflows

VSWarehouse - Variant Warehouse Server

- Archive full VCFs for Sample Cohort
 - Optimized repository infrastructure for rapid queried through millions of variants
- Centralized Genomic Data Hosting
 - Integrated with other systems
- Query Variant Warehouse
 - Have I seen this variant in my cohort?
 - Has the classification changed with new evidence in ClinVar?
 - How do I ensure consistency of CNV/variant interpretations across all users?



The screenshot displays the VSWarehouse web interface. The browser address bar shows the URL: `warehouse.goldenhelix.com/variants/Cancer%20Gene%20Panel/1/results/`. The page header includes navigation links for 'WAREHOUSE', 'Workspace', 'Query', 'Results', and 'Logout', along with the user email 'rudu@goldenhelix.com'. The main content area shows a table of 1.3 K records, with the first few rows visible:

Genomic Coordinate	Ref/Alt	Gene Names	HGVSp. (Clinically Relevant)
1:2494329	G/A	TNFRSF14	NP_003811.2:p.Val241Ile
1:11190803	C/T	MTOR	NP_004949.1:p.Glu1799Lys
1:27105927	-/G	ARID1A	NP_006006.3:p.Gly1848fs
1:27105930	G/-	ARID1A	NP_006006.3:p.Asp1850fs
1:43814978	G/A	MPL	NP_005364.1:p.Ser505Asn
1:43814980	G/A	MPL	NP_005364.1:p.Ala506Thr
1:43815007	T/A	MPL	NP_005364.1:p.Trp515Arg
1:43815007	T/C	MPL	NP_005364.1:p.Trp515Arg
1:43815008	G/C	MPL	NP_005364.1:p.Trp515Ser
1:45797504	C/G	MUTYH	NP_001121897.1:p.Gln338His
1:65332610	C/T	JAK1	NP_002218.2:p.Val310Ile
1:97770919	C/T	DPYD	NP_000101.2:p.Val732Ile
1:97981394	T/C	DPYD	NP_000101.2:p.Ile543Val
1:115252203	C/T	NRAS	NP_002515.1:p.Ala146Thr
1:115256517	T/A	NRAS	NP_002515.1:p.Ser65Cys
1:115256537	T/A	NRAS	NP_002515.1:p.Gln61His

On the right side of the interface, there is a sidebar titled '1.3 K variants' with several filter options, all of which are checked:

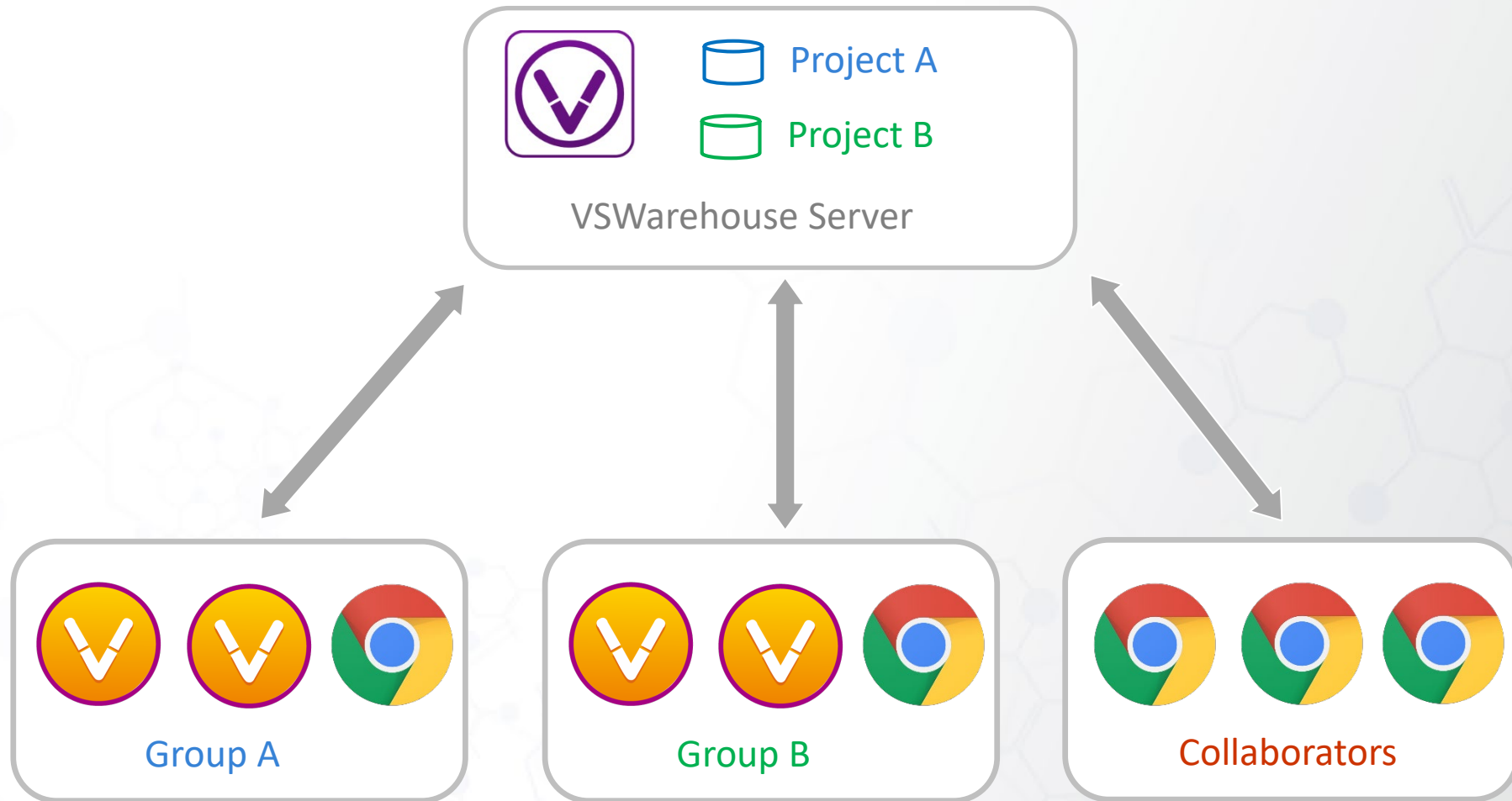
- In COSMIC? is True
- Read Depths (DP) is greater than or equal to 100.0 or Read Depths (DP) is null for Sample1
- Genotype Qualities (GQ) is greater than or equal to 80.0 or Genotype Qualities (GQ) is null for Sample1
- Effect (Combined) is either Missense or LoF

Integration with VarSeq

- From VarSeq to VSWarehouse
 - ✓ Upload full cohort data
 - ✓ Upload variant classification/interpretations
 - ✓ Upload clinical reports
- From VSWarehouse to VarSeq
 - ✓ Annotate with cohort data
 - Variant frequencies
 - ✓ Annotate with classified variants
 - ✓ Access congruent Assessment Catalogs & Reports
- VSWarehouse Browser
 - ✓ Query all genomic data
 - Reports, catalogs, projects
 - ✓ Evidence updates with ClinVar
 - ✓ Manage access permissions

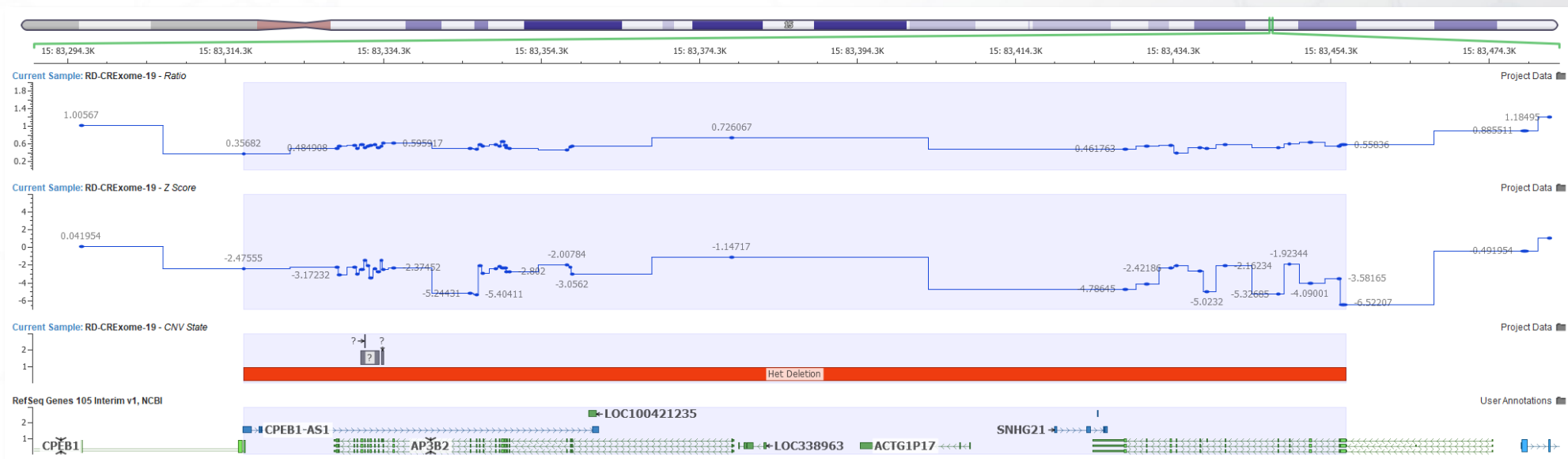


VSWarehouse – Centralized Collaboration



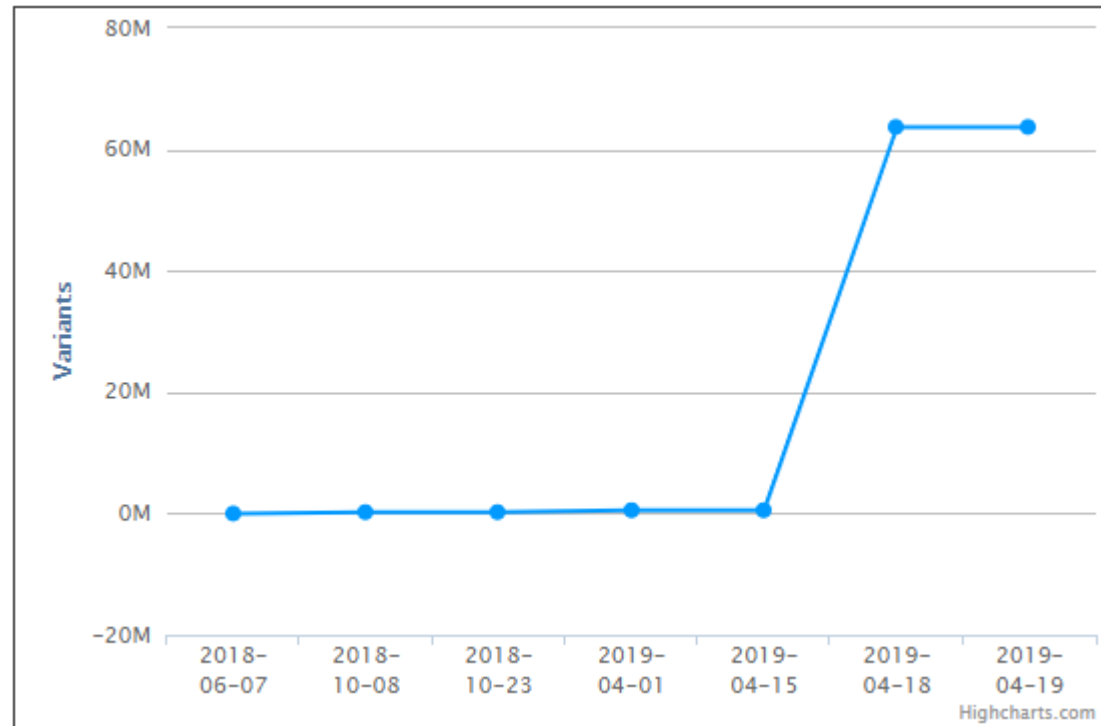
Example Project

- Part I – Exploring VSWarehouse Browser
 - General variant search
 - Filtering/query
 - Classification changes in ClinVar
- Part II – VarSeq & VSWarehouse
 - Filtering out common variants



V Warehouse®

Scalable Variant Warehouse for VarSeq



Project Demonstration >

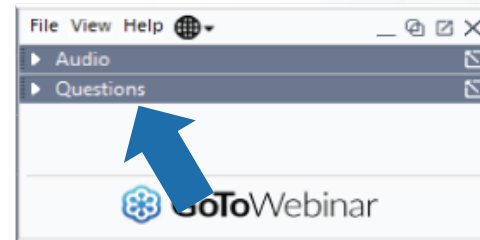
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