GOLDEN HELIX
Enabling Precision Medicine

VSWarehouse; a scalable, rapid genomic repository solution

Darby Kammeraad
Field Application Scientist Manager

CIOReview
20 Most Promising Biotech Technology Providers

Gartner
Hype Cycle for Life sciences

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Top 10 Analytics Solution Providers

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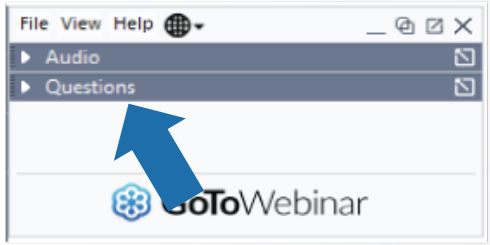

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    - Award Number R43GM128485-01
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    - Montana SMIR/STTR Matching Funds Program Grant Agreement Number 19-51-RCSBIR-005
- PI is Dr. Andreas Scherer, CEO Golden Helix.
- The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

Who Are We?

GOLDEN HELIX
Enabling Precision Medicine

Golden Helix is a global bioinformatics company founded in 1998

varSEQ®	WAREHOUSE	SNP & VARIATION SUITE
Filtering and Annotation	Variant Warehouse	CNV Analysis
ACMG Guidelines	Centralized Annotations	GWAS | Genomic Prediction
Clinical Reports	Hosted Reports	Large-N Population Studies
CNV Analysis	Sharing and Integration	RNA-Seq
Pipeline: Run Workflows		Large-N CNV-Analysis

Cited in 1,000s of Peer-Reviewed Publications

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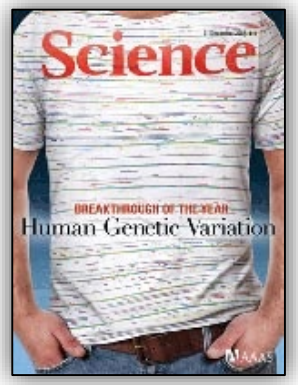

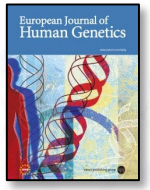

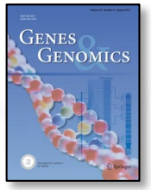

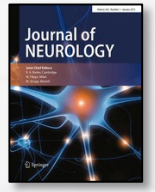

Over 400 Customers Globally

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

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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

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Gene Panel	Exome	Genome

Sequencer

Products	Bioinformatics Pipeline	Function
DNAseq (Sentieon) TNseq (Sentieon) VS-CNV	FASTQ BAM VCF	▸ Single nucleotide variation ▸ Copy number variation & loss of heterozygosity ▸ Chromosomal aberration
Annotations	Annotated VCF	▸ Public & commercial annotations to enrich genomic data sets
VarSeq VSReports VSPipeline	Clinical Report	▸ Annotate & filter ▸ Visually inspect alignments ▸ Variant prioritization ▸ Clinical assessment
VSClinical	Automated ACMG Guidelines	▸ Clinical variant interpretation in concordance with ACMG Guidelines
VSWarehouse	Data Warehousing Web-Enabled Interface + Powerful API: JSON, XML, TSV, CSV, SQL, FHIR	▸ Clinical assessment catalog ▸ Advanced data querying ▸ Versioning ▸ Interoperability ▸ Compliance with HIPPA, CLIA & CAP data discovery

VarSeq Suite

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VarSeq®

Simple

Flexible

Scalable

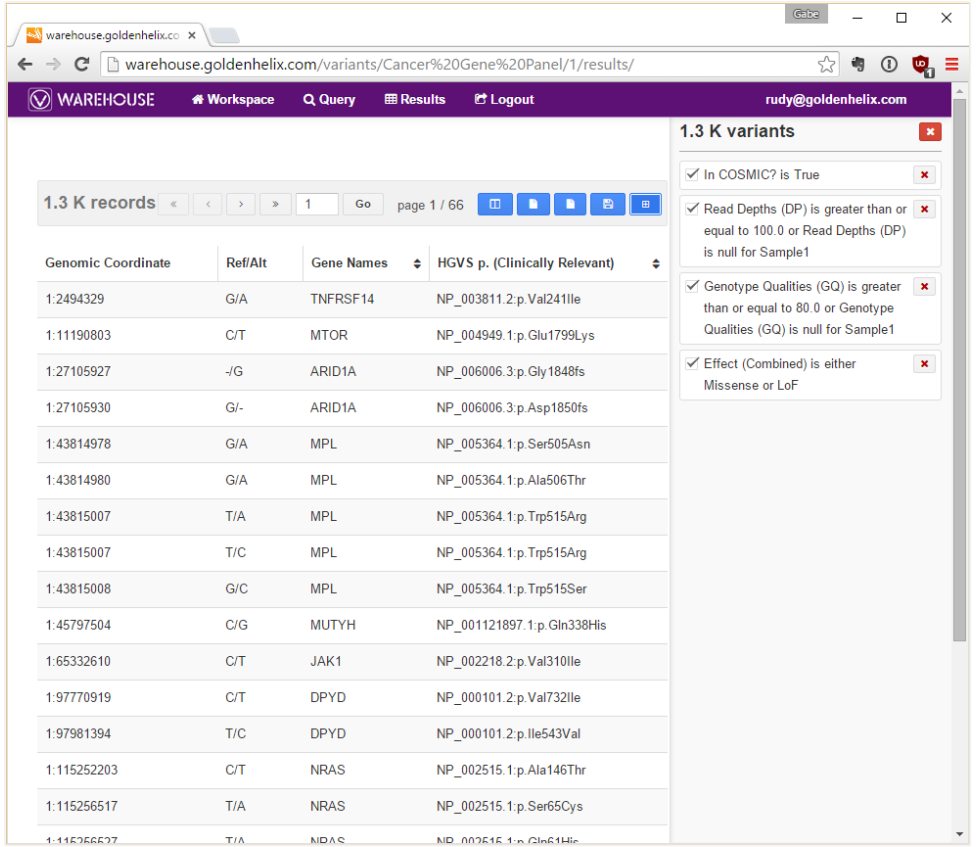
Variant annotation
filtering, and interpretation

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?



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

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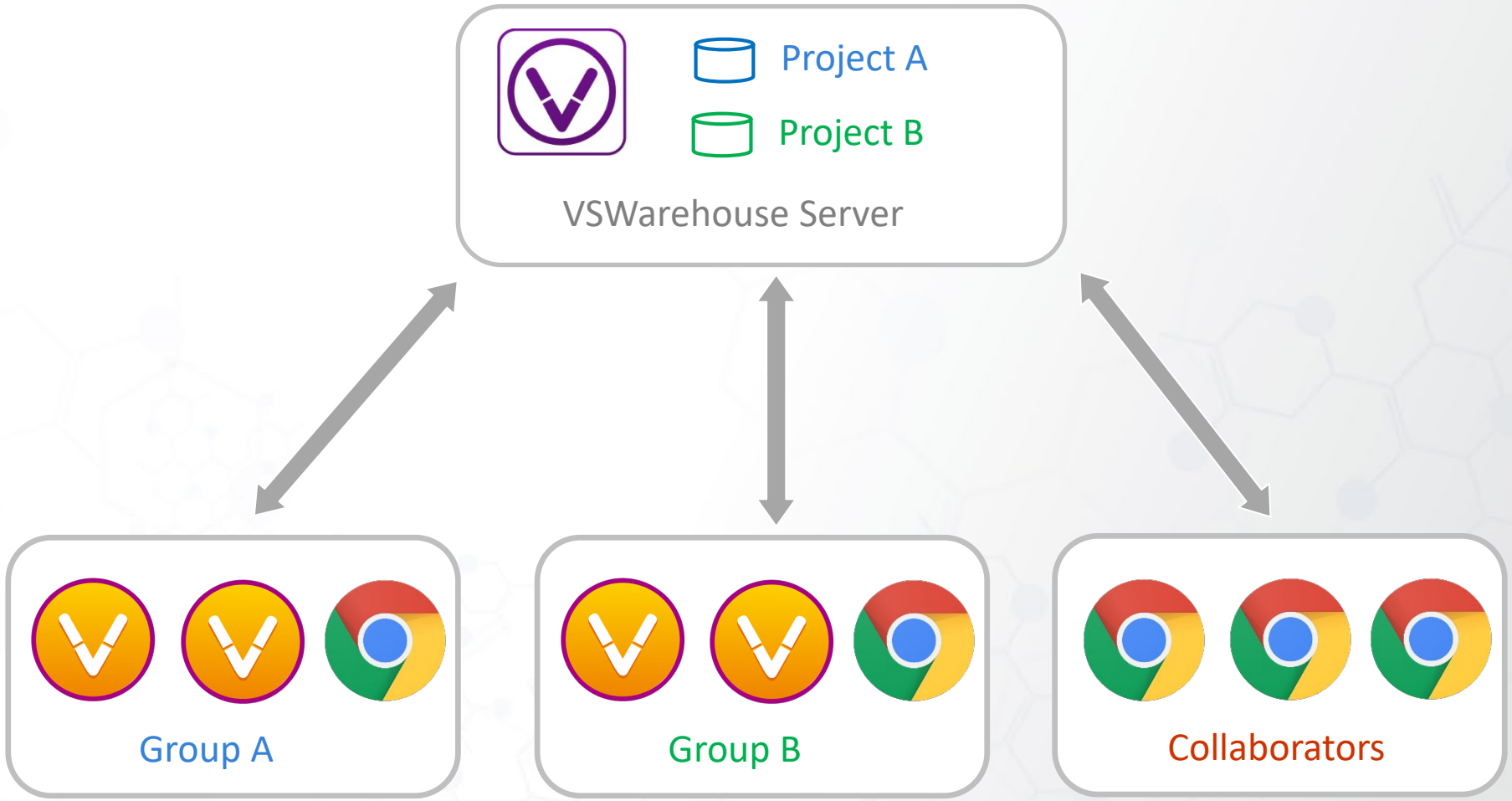
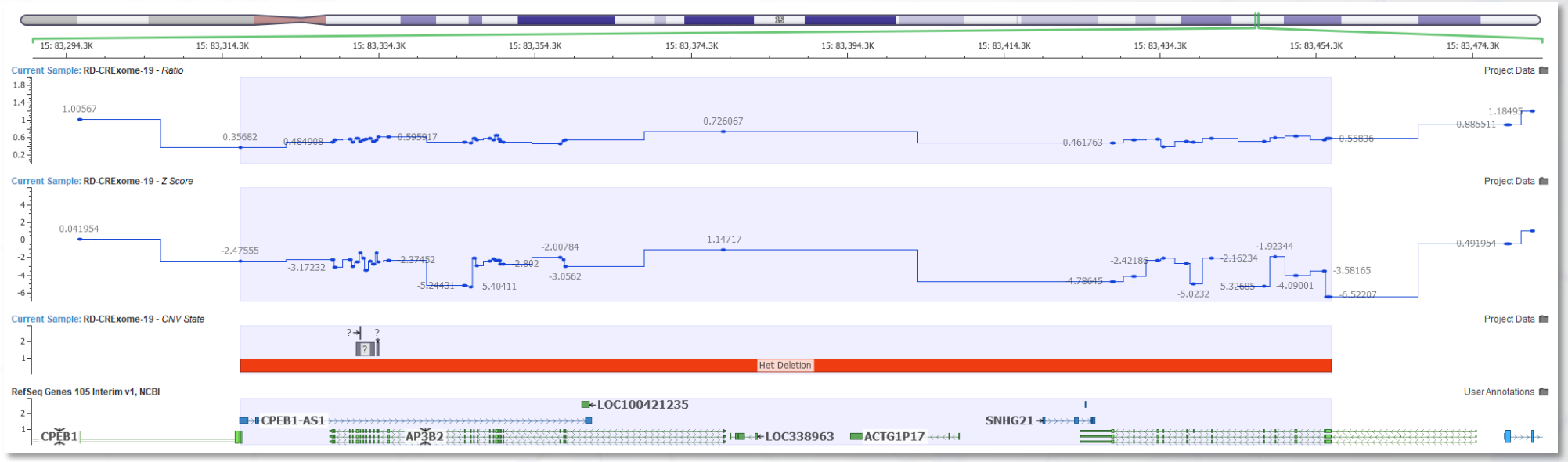


Example Project

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



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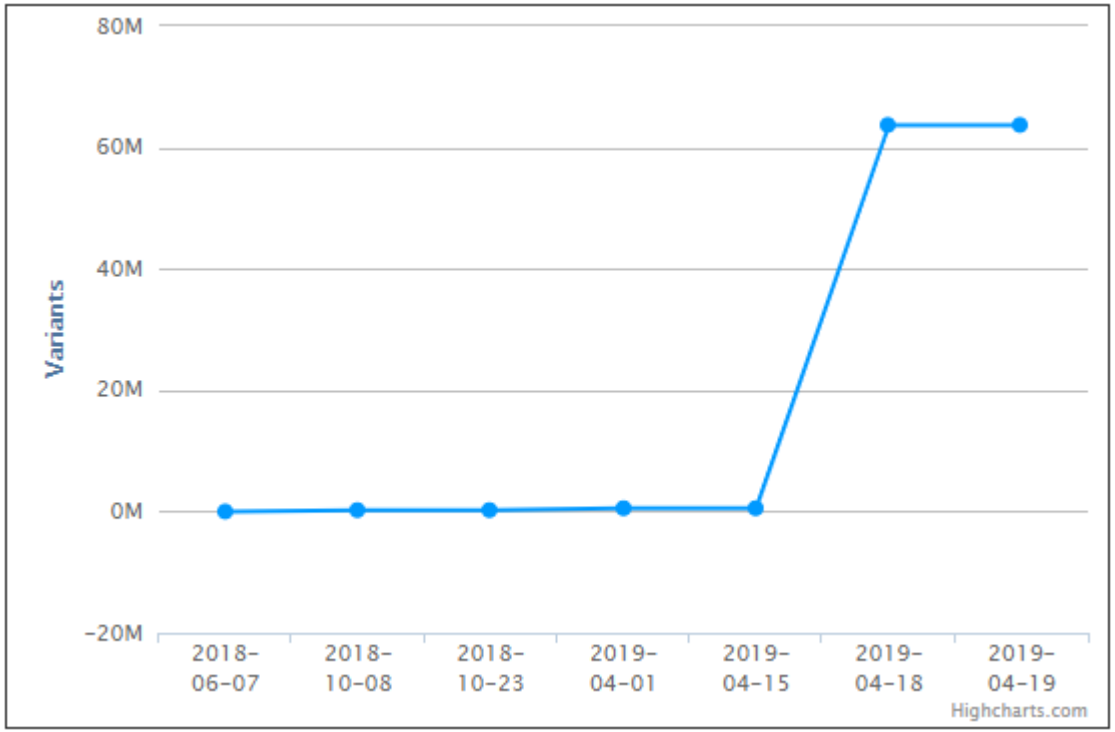
VSWarehouse®

Scalable Variant Warehouse for VarSeq



Project Demonstration >

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