Authoring Clincal Reports in VarSeq

September 23, 2015

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VP Product & Engineering

GOLDEN HELIX
Accelerating the Quest for Significance™

Agenda

1. Clinical NGS Testing Workflows
2. One Click Clinical Reporting
3. VarSeq: Clinical NGS Testing Platform
4. VSPipeline & VarSeq Viewer

Golden Helix – Who We Are

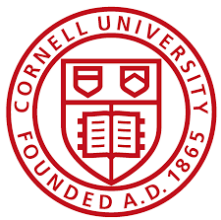
Golden Helix is a global bioinformatics company founded in 1998.

GlaxoSmithKline

We are cited in over 900 peer-reviewed publications

Our Customers

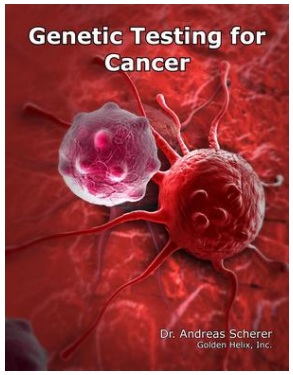
Over 200 organizations world wide, and thousands of users, trust our software.



Golden Helix – Who We Are

When you choose a Golden Helix solution, you get more than just software

- REPUTATION
- TRUST
- EXPERIENCE



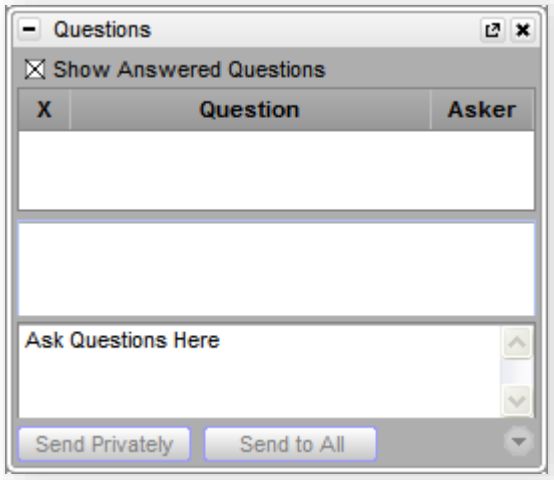
- INDUSTRY FOCUS
- THOUGHT LEADERSHIP
- COMMUNITY

- TRAINING
- SUPPORT
- RESPONSIVENESS

- TRANSPARENCY
- INNOVATION and SPEED
- CUSTOMIZATIONS

Questions during the presentation

Use the Questions pane in your GoToWebinar window



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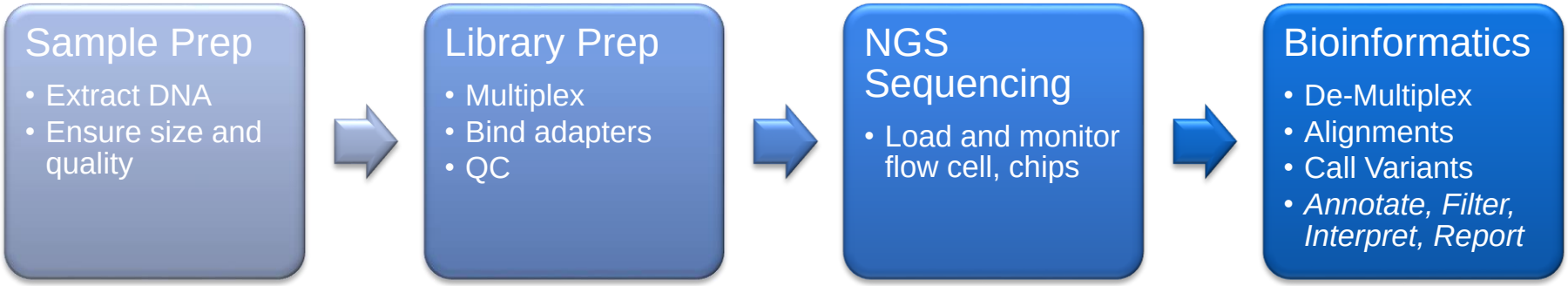
Clinical Gene Panel Tests



Annotate, Filter, Interpret, Report

- QC variants and target regions
- Annotations current & accurate
- Visualize NGS alignment, context
- Capture variant assesements, results
- Export captured and computed data

Economics

- Human hands-on-time most variable
- Lab tech and MD time highly valued
- Some analytics can be automated
- Interpretation platform optimizes for:
  - Efficient interpretation
  - Integrated with lab workflow

What is VarSeq?

- **Variant annotation, filtering and interpretation**
- **Repeatable workflows**
- **Rich visualizations with GenomeBrowse built-in**
- **Powerful GUI and command-line interfaces**

VarSeq Clinical Suite

VarSeq

- Gene panel testing
- Whole exome and whole genome analysis
- Cancer diagnostics
- Repeatable workflows
- Coverage statistics
- Multiple sample support
- Variant assesement database

VSPipeline

- Command line runner
- Integrate with your current bioinformatics pipeline
- Create repeatable clinical workflows for CLIA and CAP certified analysis
- Supports high throughput scenarios

VSReports

- Tertiary analysis to report in **one click**
- Focused and actionable data
- Modeled on ACMG guidelines
- Hereditary and cancer templates
- OMIM included

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3. VarSeq: Clinical NGS Testing Platform
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[Demonstration]

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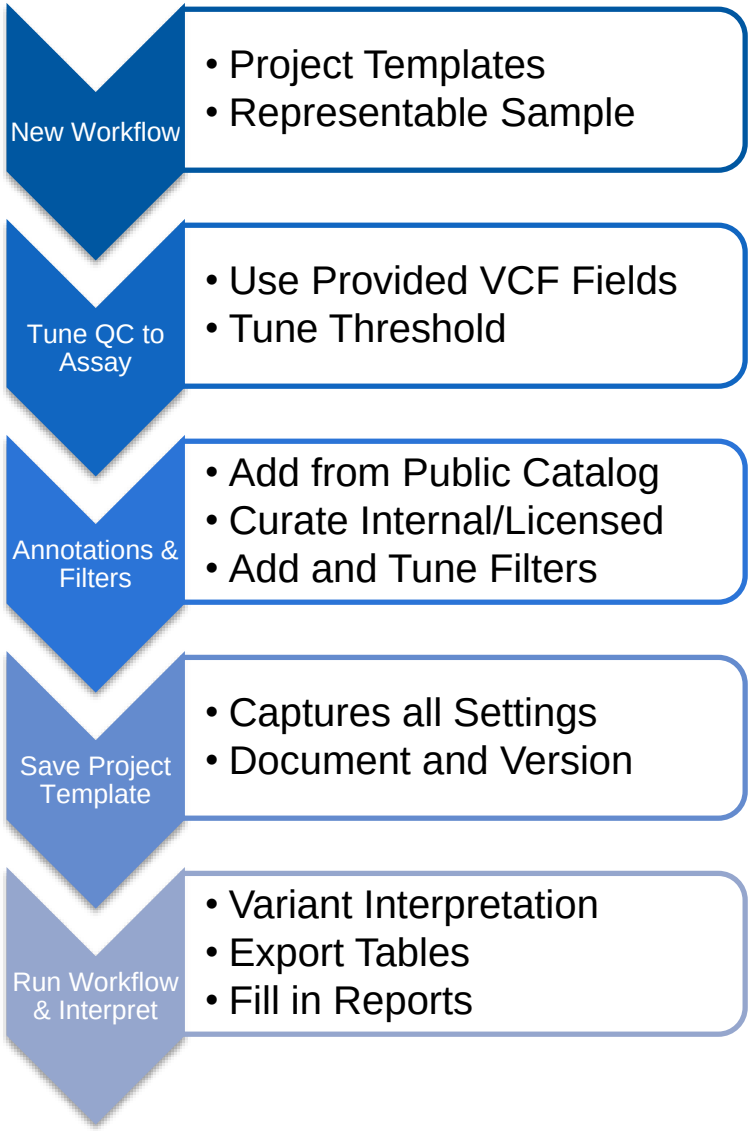
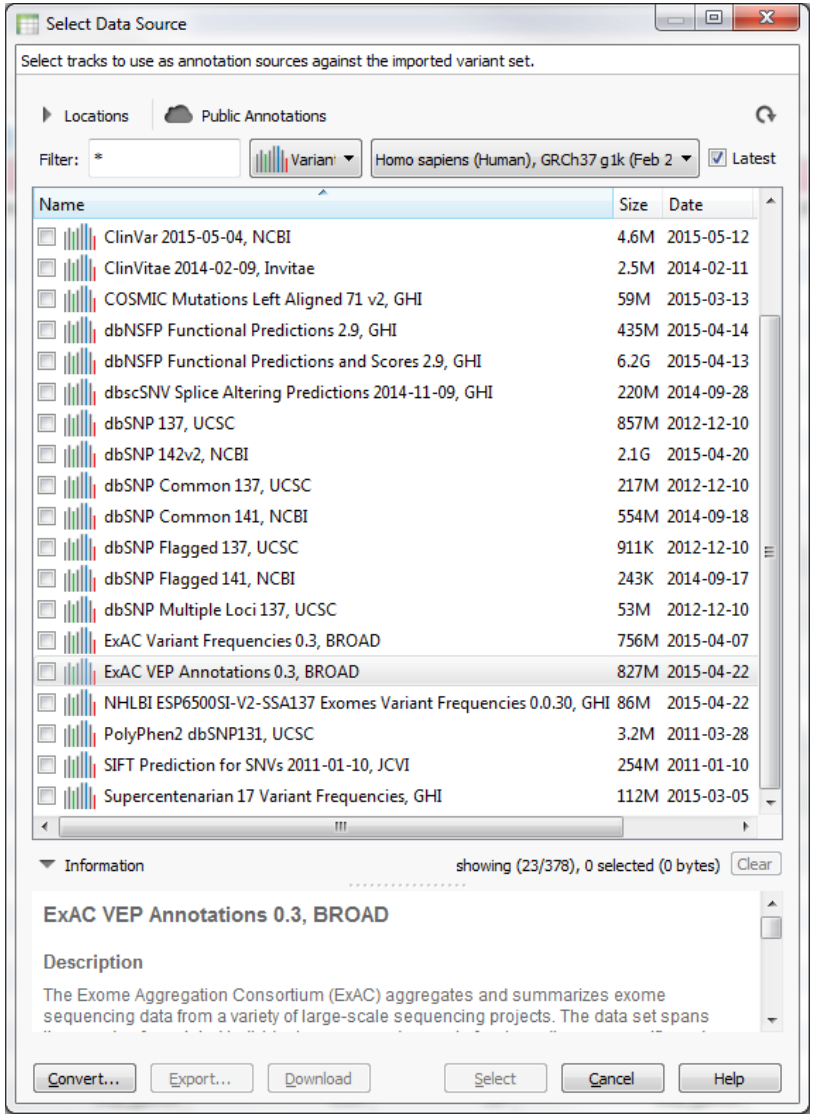
3. **VarSeq: Clinical NGS Testing Platform**
4. VSPipeline & VarSeq Viewer

Setting up a Clinical Test with VarSeq



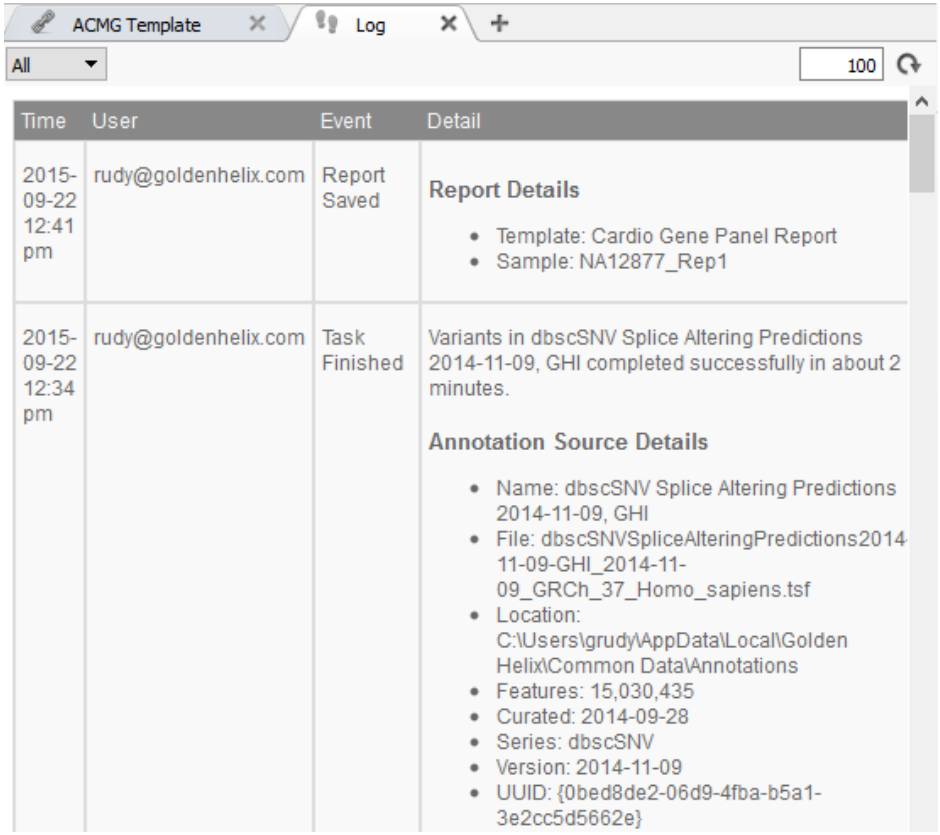
Data Curation of Annotation Sources

- **OMIM – Variant, Gene and Phenotype**
  - Bundled with Clinical Reports
  - Updated monthly
  - Rich integration of hyperlinks, references
- **VarSeq is backed by an extensive list of curated data sources**
  - 1kG Phase3 Variant Frequencies
  - ClinVar, NCBI
  - ClinVitae, Invitae
  - COSMIC
  - dbNSFP Functional Predictions
  - dbSNP
  - ExAC
  - RefSeq Genes, NCBI
  - Supercentenarian 17 Variant Frequencies
- **Your workflows lock down specific versions**



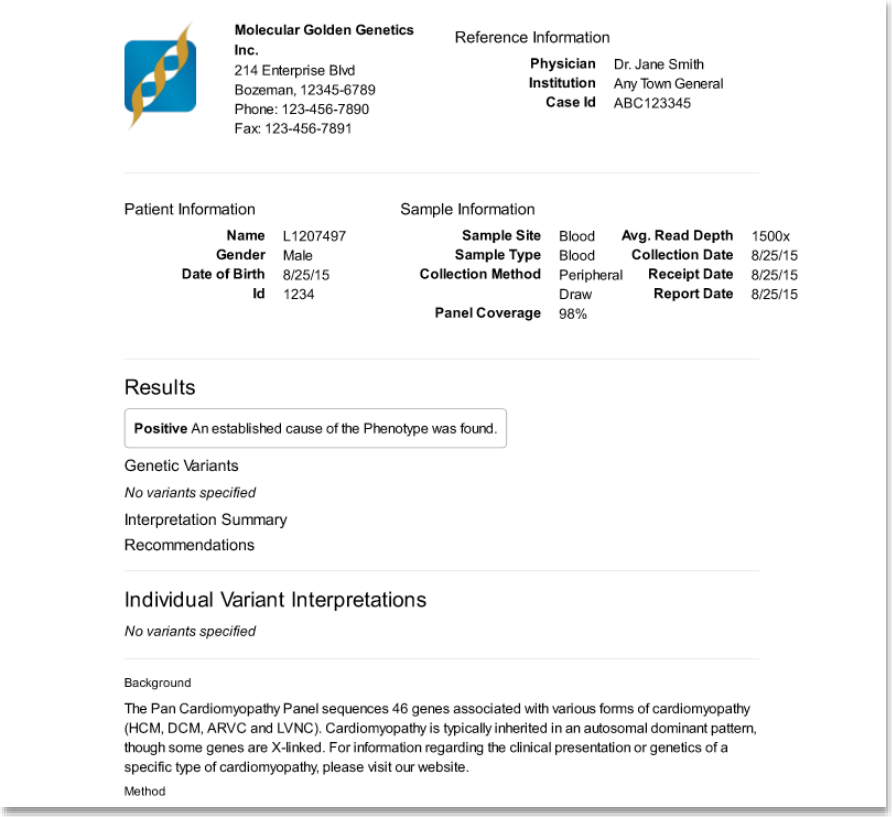
Workflows: What is it Capturing and How?

- **Save as Template**
  - Import Settings
  - Annotations and Algorithms
  - Filters (can be locked)
  - View choices (Reports, Browser etc)
- **Create New Projects**
  - Shipped “starter” templates
  - Your custom template
- **Automated and Logged**
  - Log contains what, when, whom
  - Prompts to downloaded missing sources



VSReports

- **Customize a “Report Template”**
  - ACMG Standard Germline Report
  - Configurable Global Settings
    - Logo
    - Lab Information
    - Test Description / Disclaimers
- **Customizable Sample Inputs**
  - Patient Information
  - Test Results
- **Select Variants to Include**
  - Per-variant classification
  - Interpretation prefilled from project data
- **Customizable**



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214 Enterprise Blvd
Bozeman, 12345-6789
Phone: 123-456-7890
Fax: 123-456-7891

Reference Information

Physician	Dr. Jane Smith
Institution	Any Town General
Case Id	ABC123345

Patient Information

Name	L1207497
Gender	Male
Date of Birth	8/25/15
Id	1234

Sample Information

Sample Site	Blood	**Avg. Read Depth**	1500x
Sample Type	Blood	**Collection Date**	8/25/15
Collection Method	Peripheral Draw	**Receipt Date**	8/25/15
		Report Date	8/25/15
Panel Coverage	98%		

Results

Positive An established cause of the Phenotype was found.

Genetic Variants

No variants specified

Interpretation Summary

Recommendations

Individual Variant Interpretations

No variants specified

Background

The Pan Cardiomyopathy Panel sequences 46 genes associated with various forms of cardiomyopathy (HCM, DCM, ARVC and LVNC). Cardiomyopathy is typically inherited in an autosomal dominant pattern, though some genes are X-linked. For information regarding the clinical presentation or genetics of a specific type of cardiomyopathy, please visit our website.

Method

[Demonstration]

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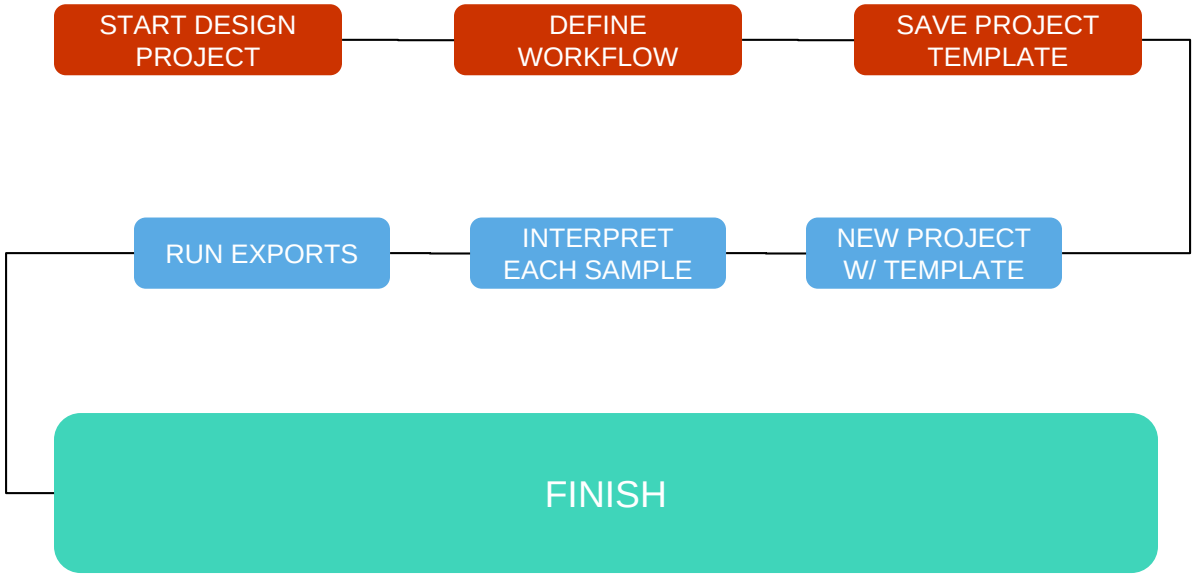
Batch Analysis Workflow

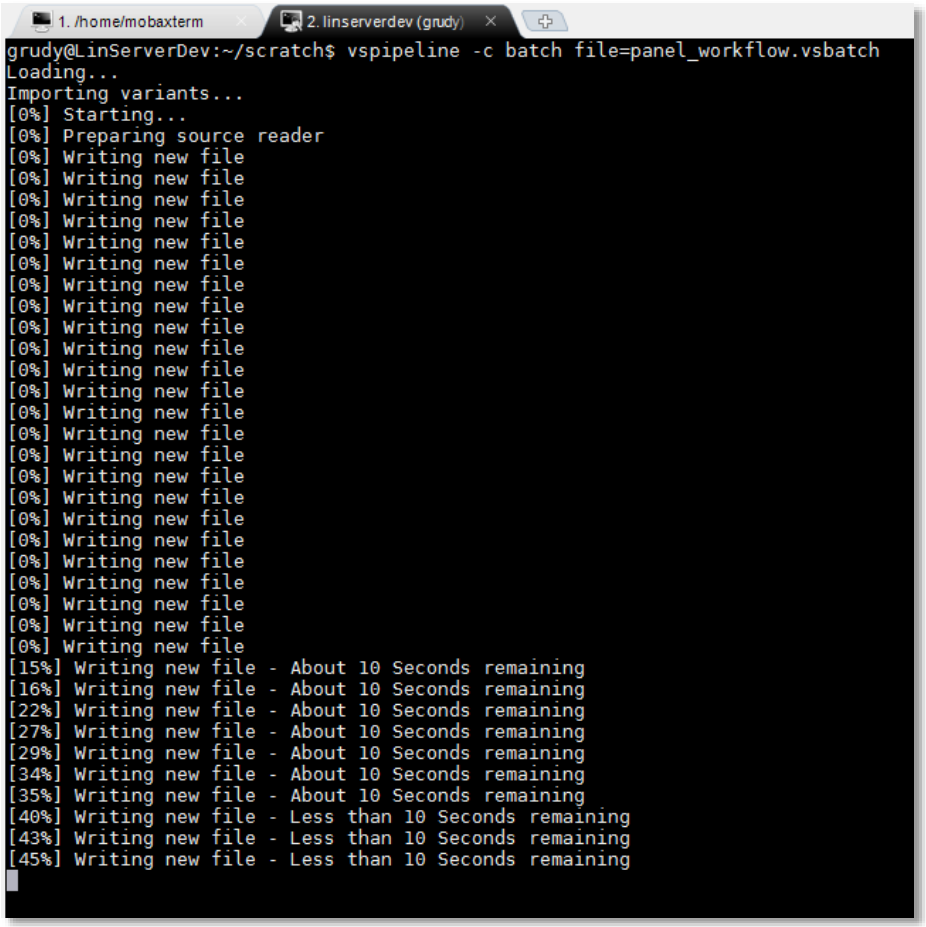
Define a Workflow that is Repeated for “Batches” of Samples



- Steps in RED are done once when designing a new workflow to be tuned to the upstream pipeline and test thresholds
- Steps in BLUE are done for each sample or set of samples that should repeat the workflow
- Steps in BLUE can be automated with **VSPIPELINE**

VSPipeline: High Throughput, Automated

- **Produce Automated "Deliverables"**
  - Filtered and annotated variant lists
  - Exported to Excel, VCF, Text
  - VarSeq Projects (openable by VarSEq, VarSeq Viewer)
- **Deploy a Project Template**
  - Support validation (like CAP/CLIA)
  - All steps logged
- **Integrated**
  - Run as part of pipeline that produces BAM/VCF files.
- **Works with VSReports!**



Available Now: VarSeq Viewer

FREE Reader of VarSeq Projects

VIEW FILTERED
View tables
Investigate results

VISUALIZE
GenomeBrowse
BAM and VCF
Save Screenshots

EXPORT
Excel Rich Reports
Text and VCF

SHARE
Relocatable Projects
Efficient Storage
Anybody can Open

VarSeq projects are a perfect preservation of an analysis, and can be opened at any time with the FREE VarSeq Viewer.

VarSeq Viewer is VarSeq without an active subscription license. It can be downloaded by anyone at anytime.

Questions or more info:

- Email info@goldenhelix.com
- Request an evaluation of the software at www.goldenhelix.com

Questions?

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