



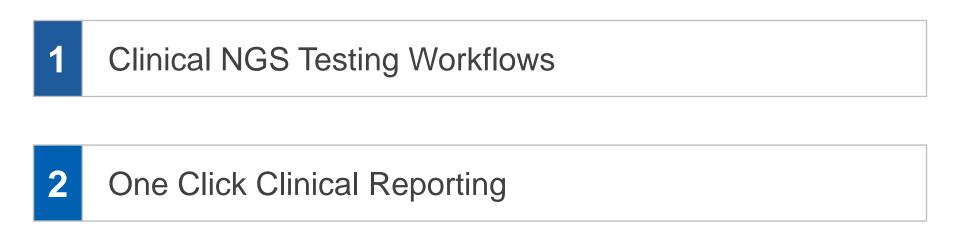
September 23, 2015

Gabe Rudy VP Product & Engineering









# **3** VarSeq: Clinical NGS Testing Platform

# 4 VSPipeline & VarSeq Viewer



Golden Helix is a global bioinformatics company founded in 1998.



#### We are cited in over 900 peer-reviewed publications





GOLDEN HELX

Accelerating the Quest for Significance



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

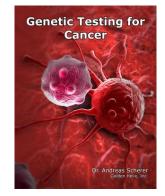




When you choose a Golden Helix solution, you get more than just software

- REPUTATION
- TRUST
- EXPERIENCE





- INDUSTRY FOCUS
- THOUGHT LEADERSHIP
- COMMUNITY

- TRAINING
- SUPPORT
- RESPONSIVENESS





- TRANSPARENCY
- INNOVATION and SPEED
- CUSTOMIZATIONS

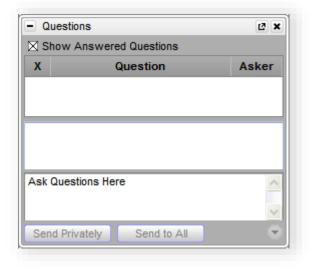






# Questions during the presentation

Use the Questions pane in your GoToWebinar window









# 1 Clinical NGS Workflows



# **3** VarSeq: Clinical NGS Testing Platform

# 4 VSPipeline & VarSeq Viewer



### **Clinical Gene Panel Tests**





- Extract DNA
- Ensure size and quality

#### Library Prep

- Multiplex
- Bind adaptersQC



• Load and monitor flow cell, chips



#### **Bioinformatics**

- De-Multiplex
- Alignments
- Call Variants
- Annotate, Filter, Interpret, Report

#### Annotate, Filter, Interpret, Report

- QC variants and target regions
- Annotations current & accurate
- Visualize NGS alignment, context
- Capture variant assessments, 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



Golden Helix





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

## VarSeq Clinical Suite



# **VORSEQ**®

#### VarSeq

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

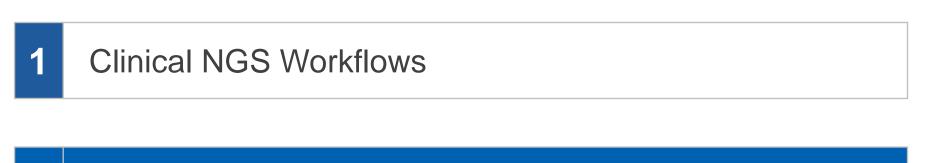
#### **VSReports**

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









2 One Click Clinical Reporting

# 3 VarSeq: Clinical NGS Testing Platform

# 4 VSPipeline & Viewer





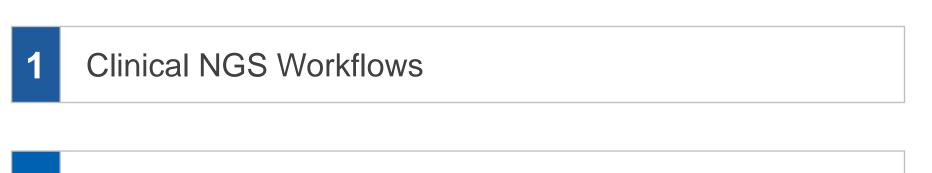


# [Demonstration]









2 One Click Clinical Reporting

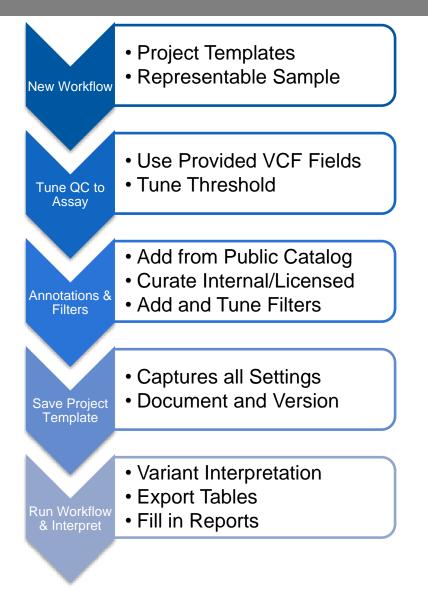
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# Setting up a Clinical Test with VarSeq







## **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
     dbSNP Frequencies - ExAC

- ClinVar, NCBI
- ClinVitae, Invitae
- COSMIC
- dbNSFP Functional Predictions
- RefSeq Genes, **NCBI**
- Supercentenarian **17** Variant Frequencies
- Your workflows lock down specific versions

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Name		*		Size	Date	*
	ClinVar 201	5-05-04, NCBI		4.6M	2015-05-12	
	ClinVitae 2	014-02-09, Invitae		2.5M	2014-02-11	
	COSMIC M	utations Left Aligned 71	v2, GHI	59M	2015-03-13	
	dbNSFP Fu	nctional Predictions 2.9,	GHI	435M	2015-04-14	
	dbNSFP Fu	nctional Predictions and	Scores 2.9, GHI	6.2G	2015-04-13	
	dbscSNV S	plice Altering Predictions	2014-11-09, GHI	220M	2014-09-28	
	dbSNP 137	UCSC		857M	2012-12-10	
	dbSNP 142	v2, NCBI		2.1G	2015-04-20	
	dbSNP Cor	nmon 137, UCSC		217M	2012-12-10	
	dbSNP Cor	nmon 141, NCBI		554M	2014-09-18	
	dbSNP Flag	jged 137, UCSC		911K	2012-12-10	Ξ
	dbSNP Flag	jged 141, NCBI		243K	2014-09-17	
	dbSNP Multiple Loci 137, UCSC			53M	2012-12-10	
	ExAC Variant Frequencies 0.3, BROAD			756M	2015-04-07	
	ExAC VEP A	Annotations 0.3, BROAD		827M	2015-04-22	
	NHLBI ESP	5500SI-V2-SSA137 Exome	s Variant Frequencies 0.0.30, GHI	86M	2015-04-22	
	PolyPhen2	dbSNP131, UCSC		3.2M	2011-03-28	
	SIFT Predic	tion for SNVs 2011-01-10	, JCVI	254M	2011-01-10	
	Supercente	narian 17 Variant Freque	ncies, GHI	112M	2015-03-05	-
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Desc	ription					
The E	xome Aggreg		c) aggregates and summarizes			
sequ	encing data fr	om a variety of large-sca	ale sequencing projects. The da	ta set	spans	-



# 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

All 👻 100				
Time	User	Event	Detail	
2015- 09-22 12:41 pm	rudy@goldenhelix.com	Report Saved	Report Details <ul> <li>Template: Cardio Gene Panel Report</li> <li>Sample: NA12877_Rep1</li> </ul>	
2015- 09-22 12:34 pm	rudy@goldenhelix.com	Task Finished	Variants in dbscSNV Splice Altering Predictions 2014-11-09, GHI completed successfully in about 2 minutes. Annotation Source Details Name: dbscSNV Splice Altering Predictions 2014-11-09, GHI File: dbscSNVSpliceAlteringPredictions2014 11-09-GHI_2014-11- 09_GRCh_37_Homo_sapiens.tsf Location: C:\Users\grudy\AppData\Local\Golden Helix\Common Data\Annotations Features: 15,030,435 Curated: 2014-09-28 Series: dbscSNV Version: 2014-11-09 UUID: {0bed8de2-06d9-4fba-b5a1- 3e2cc5d5662e}	



## **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

Accelerating the Quest for Significa

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<b>F</b>	Molecular Golden Genetics Inc. 214 Enterprise Blvd Bozeman, 12345-6789 Phone: 123-456-7890 Fax: 123-456-7891		Reference	Reference Information Physician Dr. Jane Smith Institution Any Town General Case Id ABC123345			
Patient Inform	nation		Sample Information				
	Name Gender of Birth Id	L1207497 Male 8/25/15 1234	Sample Situ Sample Type Collection Method Panel Coverage	e Blood d Periphe Draw		vg. Read Depth Collection Date Receipt Date Report Date	1500x 8/25/15 8/25/15 8/25/15
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specific type of cardiomyopathy, please visit our website

Method



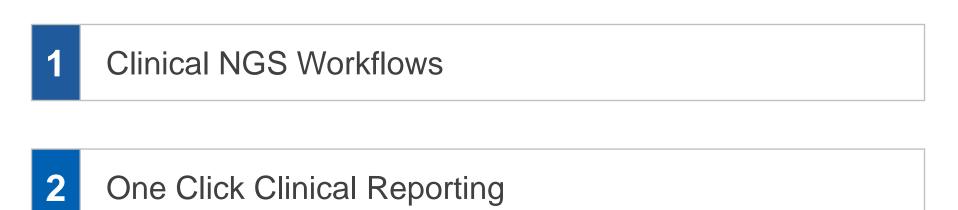


# [Demonstration]









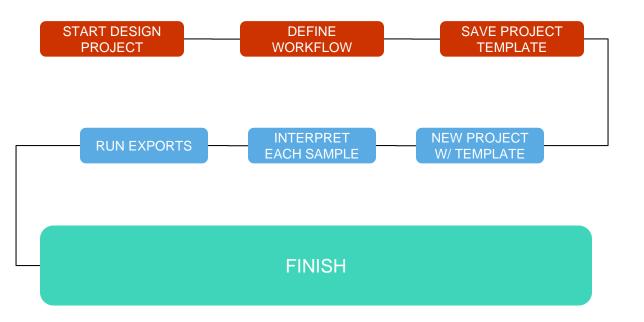
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## 4 VSPipeline & VarSeq Viewer





#### 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**



#### Produce Automated "Deliverables"

- Filtered and annotated variant lists
- Exported to Excel, VCF, Text
- VarSeq Projects (openable by VarSEq, VarSeq Viewer)

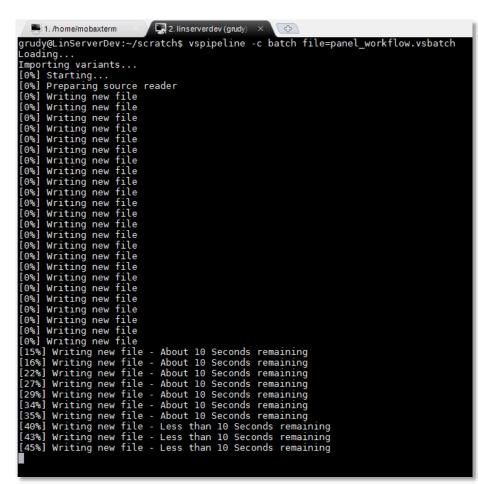
#### Deploy a Project Template

- 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



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.

Anybody can Open







# Questions or more info:

- Email info@goldenhelix.com
- Request an evaluation of the software at <u>www.goldenhelix.com</u>









# **Questions?**

Use the Questions pane in your GoToWebinar window

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