Agenda

1. Clinical NGS Testing Workflows

2. One Click Clinical Reporting

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.
Our Customers

Over 200 organizations worldwide, 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
Agenda

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

Sample Prep
- Extract DNA
- Ensure size and quality

Library Prep
- Multiplex
- Bind adapters
- QC

NGS Sequencing
- 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
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

<table>
<thead>
<tr>
<th><strong>VarSeq</strong></th>
<th><strong>VSPipeline</strong></th>
<th><strong>VSReports</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>- Gene panel testing</td>
<td>- Command line runner</td>
<td>- Tertiary analysis to report in <strong>one click</strong></td>
</tr>
<tr>
<td>- Whole exome and whole genome analysis</td>
<td>- Integrate with your current bioinformatics pipeline</td>
<td>- Focused and actionable data</td>
</tr>
<tr>
<td>- Cancer diagnostics</td>
<td>- Create repeatable clinical workflows for CLIA and CAP certified analysis</td>
<td>- Modeled on ACMG guidelines</td>
</tr>
<tr>
<td>- Repeatable workflows</td>
<td>- Supports high throughput scenarios</td>
<td>- Hereditary and cancer templates</td>
</tr>
<tr>
<td>- Coverage statistics</td>
<td></td>
<td>- OMIM included</td>
</tr>
<tr>
<td>- Multiple sample support</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Variant assessment database</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Agenda

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

[Demonstration]
<table>
<thead>
<tr>
<th></th>
<th>Agenda</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Clinical NGS Workflows</td>
</tr>
<tr>
<td>2</td>
<td>One Click Clinical Reporting</td>
</tr>
<tr>
<td>3</td>
<td>VarSeq: Clinical NGS Testing Platform</td>
</tr>
<tr>
<td>4</td>
<td>VSPipeline &amp; VarSeq Viewer</td>
</tr>
</tbody>
</table>
Setting up a Clinical Test with VarSeq

New Workflow
- Project Templates
- Representable Sample

Tune QC to Assay
- Use Provided VCF Fields
- Tune Threshold

Annotations & Filters
- Add from Public Catalog
- Curate Internal/Licensed
- Add and Tune Filters

Save Project Template
- Captures all Settings
- Document and Version

Run Workflow & Interpret
- Variant Interpretation
- Export Tables
- Fill in Reports
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
[Demonstration]
<table>
<thead>
<tr>
<th></th>
<th>Agenda</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Clinical NGS Workflows</td>
</tr>
<tr>
<td>2</td>
<td>One Click Clinical Reporting</td>
</tr>
<tr>
<td>3</td>
<td>VarSeq: Clinical NGS Testing Platform</td>
</tr>
<tr>
<td>4</td>
<td>VSPipeline &amp; VarSeq Viewer</td>
</tr>
</tbody>
</table>
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!**
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.
Questions or more info:

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

Use the Questions pane in your GoToWebinar window.