Getting Started with Golden Helix Varseq: The VarSeq User Experience

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Dr. Bryce Christensen
Director of Services

GOLDEN HELIX
Accelerating the Quest for Significance™

Questions during the presentation

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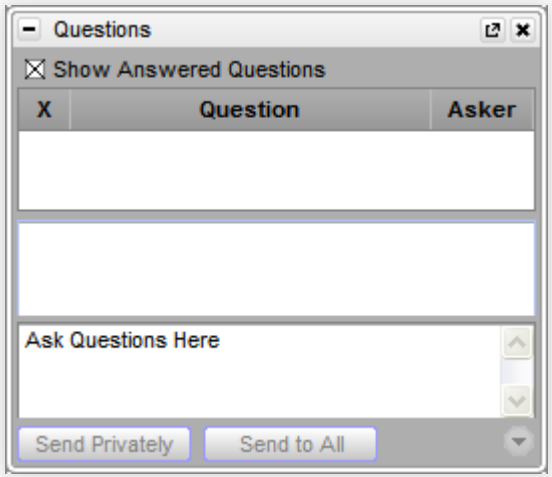


Agenda

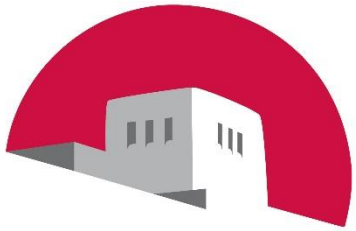
1. VarSeq Background
2. Gene Panel Analysis Demonstration
3. Family Trio Exome Analysis Demonstration
4. VarSeq Present and Future

Announcing VarSeq

- Recording available at goldenhelix.com
- Golden Helix founded in 1998
- Work on VarSeq revealed in 2013
- VarSeq built on mature technology

Stakeholders

- Jason Byars
- David Gokhale
- Kelly Eggleton
- Bruno Ping
- Cristian Ionescu-Zanetti
- Reece Hart
- Ken Kaufman
- Sam Strom
- Jeff Moore
- Jeff Rosenfeld
- Scott Ness



UNM

Liverpool Women's NHS Foundation Trust

FLUXION

UCLA Health

Royal Surrey County Hospital NHS Foundation Trust

Cincinnati Children's®

ILLINOIS
UNIVERSITY OF ILLINOIS AT URBANA-CHAMPAIGN

Demonstration: Cancer Panel

- Panels target frequently mutated regions in known human cancer genes
- Variants in certain genes may be useful to identify tumor subtypes, aiding in patient diagnosis
- Some variants may be “actionable,” or informative for patient treatment decisions

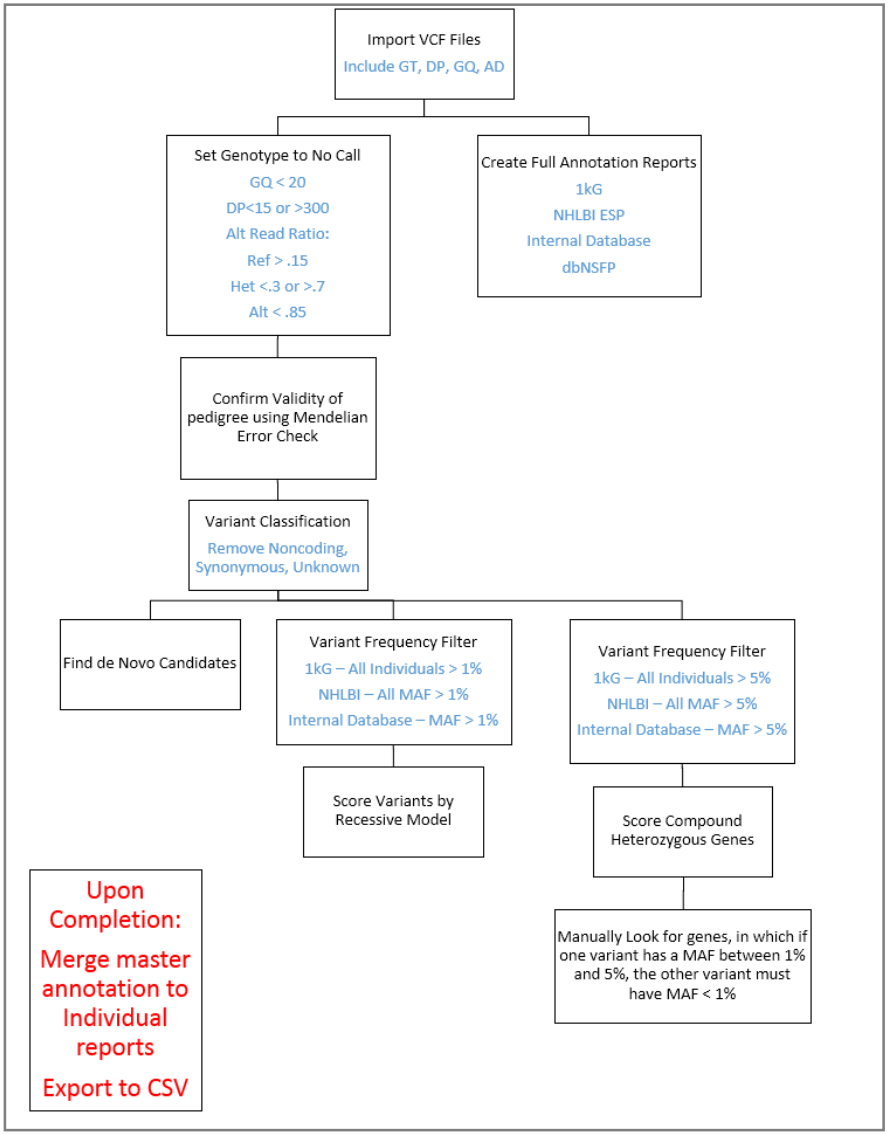
Cancer Panel Demonstration

- **Confirm coverage of all targeted hotspots**
  - Prepare report on low coverage areas
- **Evaluate variants present at targeted hotspots**
  - Review sequence quality and make report
- **Evaluate any observed mutations not in hotspot list**
  - Review and make report
- **Update analysis template if necessary, and apply to additional samples**

[Demonstration]

Trio Workflow

- **Based on workflow developed by an SVS user**
- **Similar workflow required days per case using Excel and manual processing**
- **In SVS, workflow was simplified to require 2 hours or less**
  - Many clicks and manual steps
- **VarSeq**
  - Once template is created, about 5 minutes to apply to each new trio!

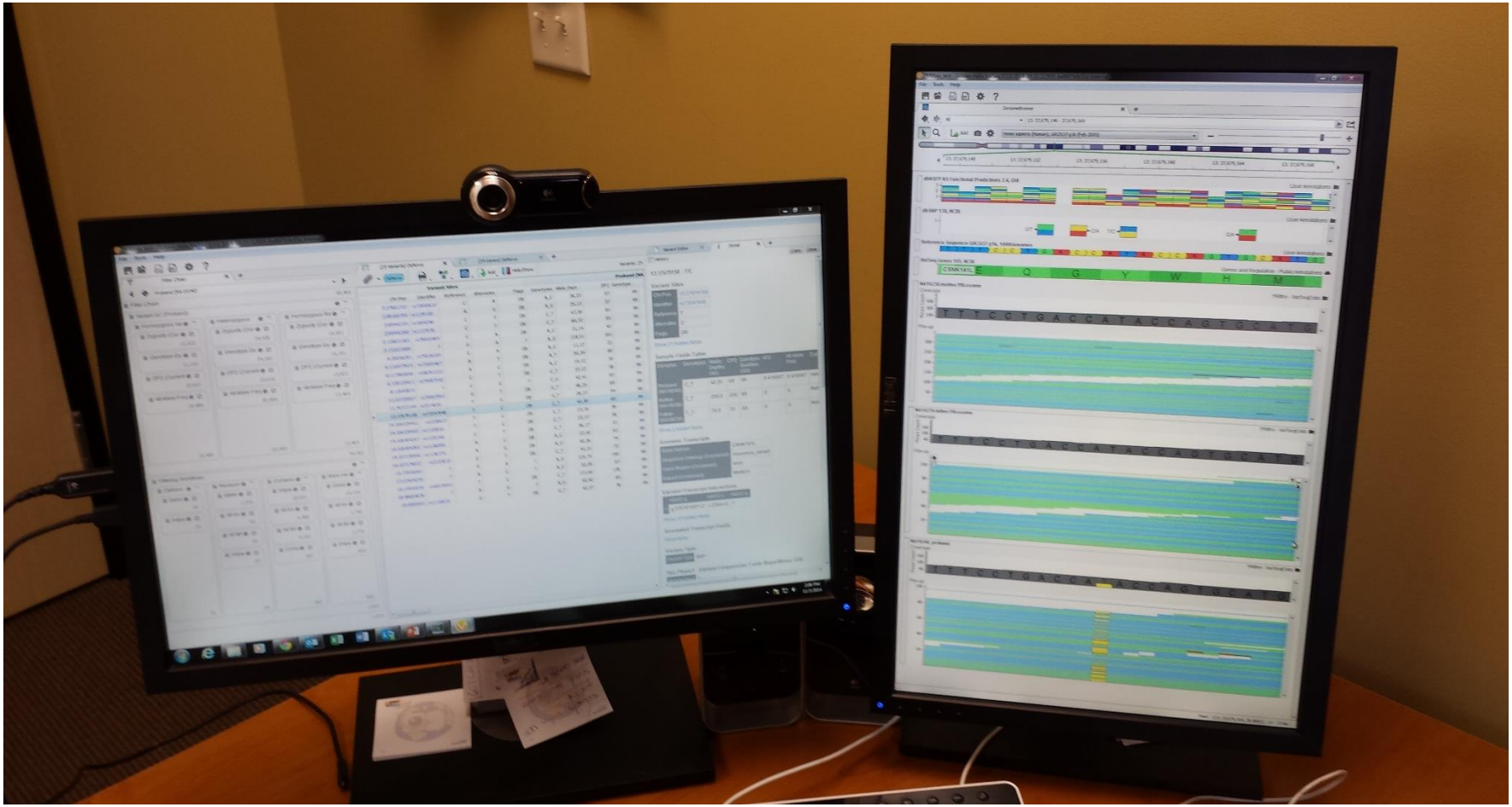


[Demonstration]

Screen Configuration in VarSeq

- **All VarSeq screen regions are modular**
  - Filter Chain
  - GenomeBrowse
  - Details
  - Table
  - Web Browser
  - Evernote
- **Can be independently moved, layered in tabs, and/or re-sized**
- **Allows workspace customization that simply isn't possible in browser-based applications**

Layout Flexibility: Bryce's Workstation



VarSeq Today

- Full featured variant annotation and filtering tool
- Simple and intuitive interface
- Fast and responsive
- Enables repeatable processes
- Gene panels, exomes
- Based on mature technology stack
- Simple licensing model

The Future of VarSeq

- **VarSeq will continue to improve**
  - Responsive release cycle
  - We want your input!

- **Features coming soon**
  - Ability to save variants to an internal database for use in future filtering
  - Enhanced project and variant note taking
  - Filtering based on custom gene lists
  - Sample level QC statistics
  - Ability to be part of existing annotation pipeline in server environments
  - Extensive algorithmic customizations

Questions or more info:

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

Questions?

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