varSEQ®

VSReports & VSPipeline

Using Clinical Reports as Part of a Gene Panel Pipeline

July 13, 2016

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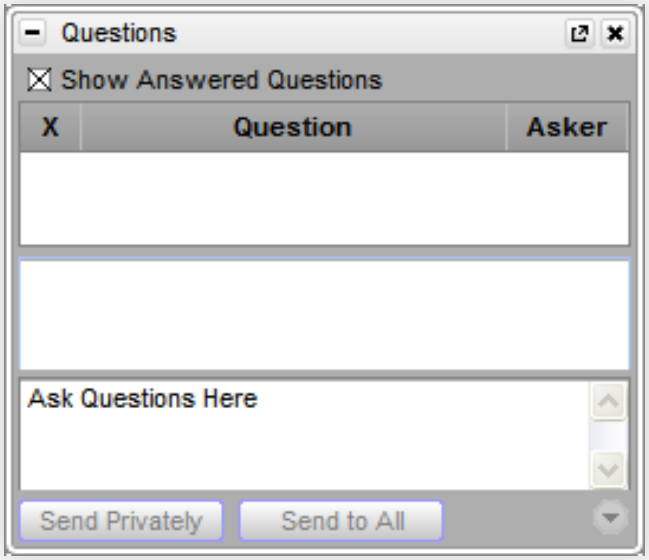
Field Application Scientist

GOLDEN HELIX

Accelerating the Quest for Significance™

Questions During The Presentation

Use the Questions pane in your GoToWebinar window



Agenda

1. Overview Golden Helix
2. VSPipeline
3. DEMO
4. Q & A

Golden Helix – Who We Are

Golden Helix is a global bioinformatics company founded in 1998.

gsk GlaxoSmithKline

SNP & VARIATION SUITE

GWAS
Genomic Prediction
Large-N-Population Studies
RNA-Seq
CNV-Analysis

varSEQ®

Filtering and Annotation
Clinical Reports
Pipeline
Data Warehousing

Over 300 customers globally

SickKids
THE HOSPITAL FOR SICK CHILDREN

Cincinnati Children's
Hospital Medical Center

The Children's Hospital of Philadelphia

Stanford University

UBC

Ucla

Yale

The Feinstein Institute for Medical Research
North Shore-Long Island Jewish Health System

North Shore LIJ

PREVENTION GENETICS
DISEASE PREVENTION THROUGH GENETIC TESTING

MAYO CLINIC

LINEAGEN

MOFFITT CANCER CENTER

Dartmouth-Hitchcock
NORRIS COTTON CANCER CENTER

BAYER

Lilly

abbvie

TEVA

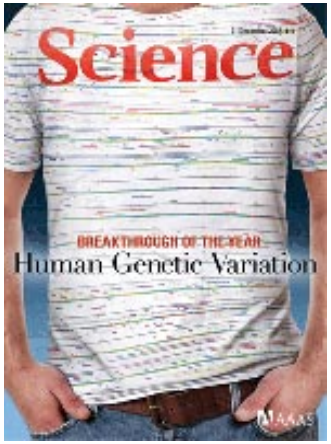
NATIONAL CANCER INSTITUTE

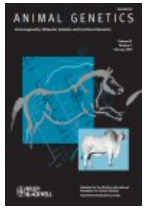
USDA
USDA Agricultural Research Service

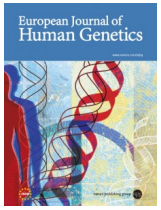
FDA

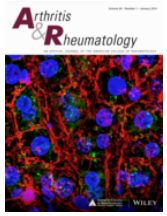
GOLDEN HELIX
Accelerating the Quest for Significance™

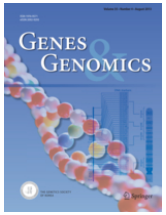
Cited in over 1000 peer-reviewed publications

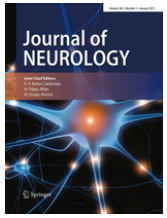








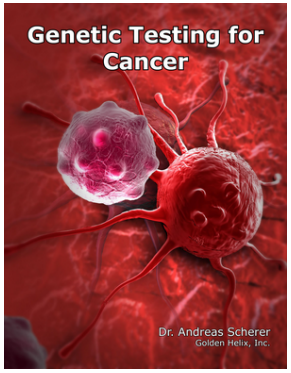




Golden Helix – Who We Are

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

- REPUTATION
- TRUST
- EXPERIENCE



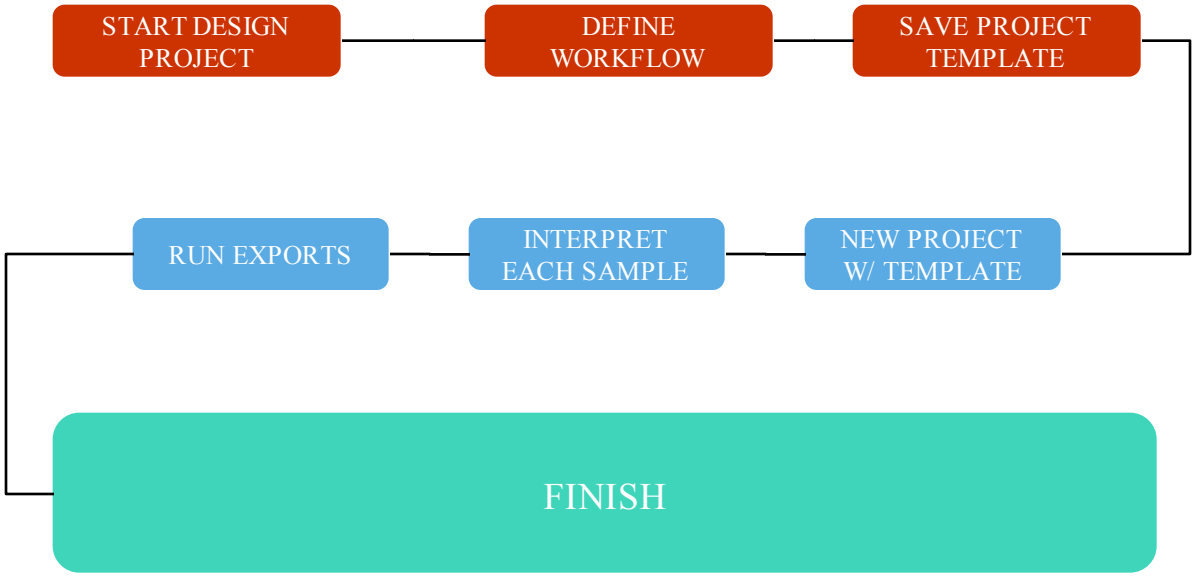
- INDUSTRY FOCUS
- THOUGHT LEADERSHIP
- COMMUNITY

- TRAINING
- SUPPORT
- RESPONSIVENESS

- TRANSPARENCY
- INNOVATION and SPEED
- CUSTOMIZATIONS

Batch Analysis Workflow

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

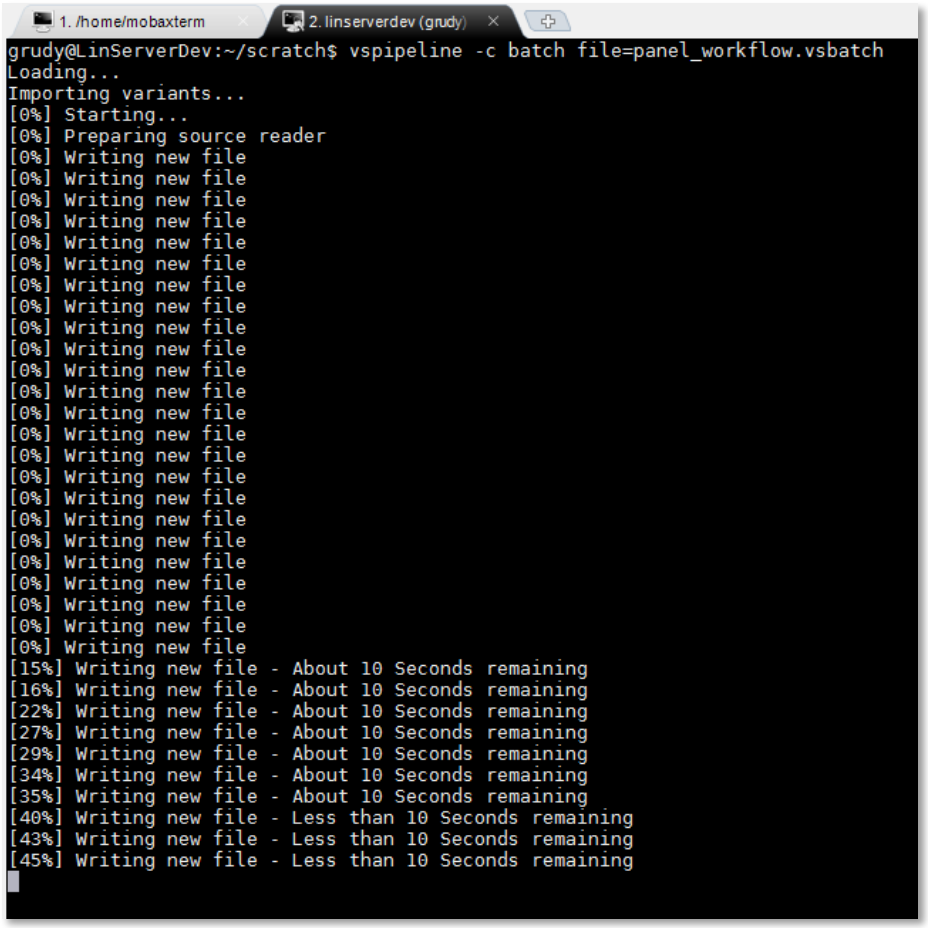


Design and Repeat

- 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
  - Clinical Report
  - 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.

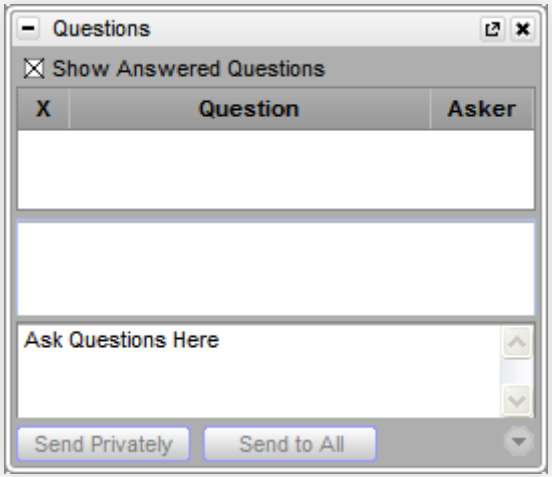


Demo

- Illumina MiniSeq TruSight Cancer Panel
- Lymphoma Association
- Onocgenes
- Tumor Suppressor Genes
- BAM and VCF files available

Questions or More Info

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

Questions?

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