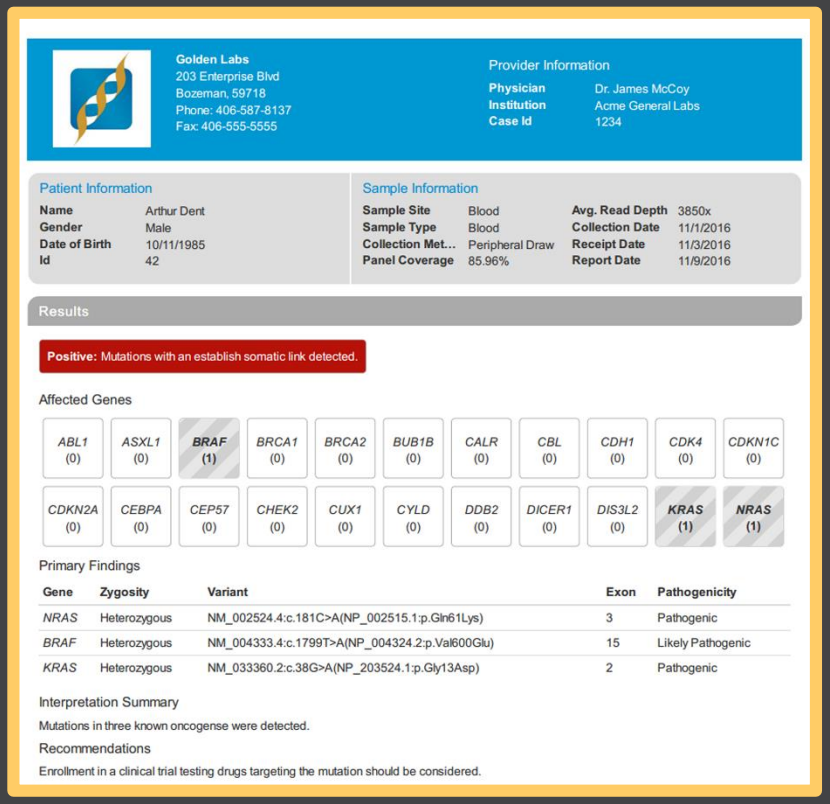
Clinical Reporting Made Easy



Golden Labs
203 Enterprise Blvd
Bozeman, 59718
Phone: 406-587-8137
Fax: 406-555-5555

Provider Information

Physician	Dr. James McCoy
Institution	Acme General Labs
Case Id	1234

Patient Information

Name	Arthur Dent
Gender	Male
Date of Birth	10/11/1985
Id	42

Sample Information

Sample Site	Blood	**Avg. Read Depth**	3850x
Sample Type	Blood	**Collection Date**	11/1/2016
Collection Met…	Peripheral Draw	**Receipt Date**	11/3/2016
Panel Coverage	85.96%	**Report Date**	11/9/2016

Results

Positive: Mutations with an establish somatic link detected.

Affected Genes

ABL1 (0)	*ASXL1* (0)	***BRAF*** **(1)**	*BRCA1* (0)	*BRCA2* (0)	*BUB1B* (0)	*CALR* (0)	*CBL* (0)	*CDH1* (0)	*CDK4* (0)	*CDKN1C* (0)
CDKN2A (0)	*CEBPA* (0)	*CEP57* (0)	*CHEK2* (0)	*CUX1* (0)	*CYLD* (0)	*DDB2* (0)	*DICER1* (0)	*DIS3L2* (0)	***KRAS*** **(1)**	***NRAS*** **(1)**

Primary Findings

Gene	Zygosity	Variant	Exon	Pathogenicity
NRAS	Heterozygous	NM_002524.4:c.181C>A(NP_002515.1:p.Gln61Lys)	3	Pathogenic
BRAF	Heterozygous	NM_004333.4:c.1799T>A(NP_004324.2:p.Val600Glu)	15	Likely Pathogenic
KRAS	Heterozygous	NM_033360.2:c.38G>A(NP_203524.1:p.Gly13Asp)	2	Pathogenic

Interpretation Summary

Mutations in three known oncogense were detected.

Recommendations

Enrollment in a clinical trial testing drugs targeting the mutation should be considered.

February 15th, 2017

Steven Hystad
Field Application Scientist

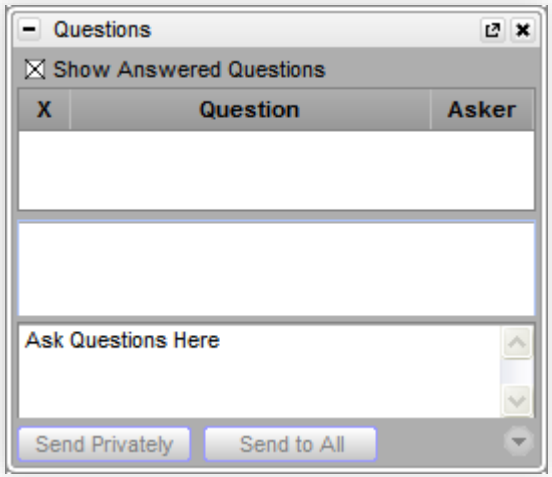
GOLDEN HELIX
Accelerating the Quest for Significance™

Agenda

1. Overview Golden Helix
2. Value of Clinical Reporting in VarSeq
3. Live Demo
4. Q&A

Questions during the presentation

Use the Questions pane in your GoToWebinar window



Golden Helix – Who We Are

Golden Helix is a global bioinformatics company founded in 1998.

GlaxoSmithKline

varSEQ®

- Filtering and Annotation
- Single Sample CNV-Analysis
- Clinical Reports
- Pipeline: Run Workflows

VARIANT WAREHOUSE

- Variant Warehouse
- Centralized Annotations
- Hosted Reports
- Sharing and Integration

SNP & VARIATION SUITE

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

Over 350 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

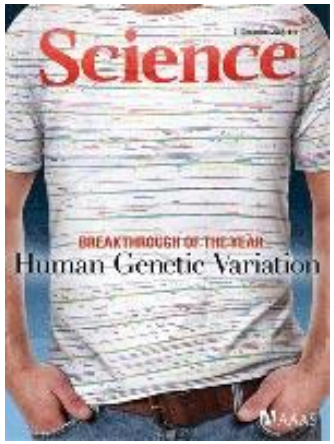
NATIONAL CANCER INSTITUTE

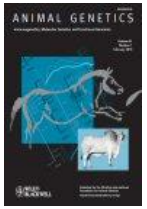
USDA
USDA Agricultural Research Service

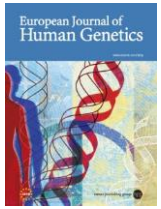
FDA

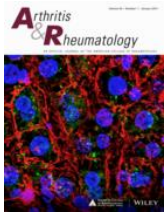
GOLDEN HELIX
Accelerating the Quest for Significance™

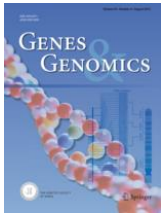
Cited in over 1000 peer-reviewed publications







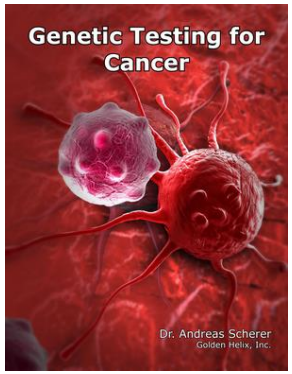




Golden Helix – Who We Are

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

VSReports


GOLDEN HELIX

Patient Information

Name	Gender	Date of Birth	Id
Jane Doe	Female	November 10, 2010	1234

Mother Information

Name	Date of Birth	Id
Jane Doe	September 28, 1988	1235

Father Information

Name	Date of Birth	Id
John Doe	November 10, 1988	1236

Reference Information

Physician	Institution	Case Id
Dr. Hystad	Golden Labs	GL-1234

Sample Information

Sample Site	Sample Type	Collection Method	Collection Date	Receipt Date	Report Date
Blood	Blood	Venipuncture	January 30, 2017	January 30, 2017	January 31, 2017

Results - Positive

Mutations with an established link detected.

Primary Findings

Gene	Exon	Variant	Zygosity	Pathogenicity
LPIN2	20	NM_014646.2:c.*3C>T	Heterozygous	Uncertain Significance

Interpretation Summary

Patient possess a a single Pathogenic de novo variants in the LPIN2 gene.

Recommendations

The variant could be associated with Chronic Recurrent Multifocal Osteomyelitis (CRMO).Treatment of CRMO typically involves administration of nonsteroidal anti-inflammatory drugs (NSAIDs) and physical therapy to avoid disuse atrophy of muscles and contractures.

Incidental Findings

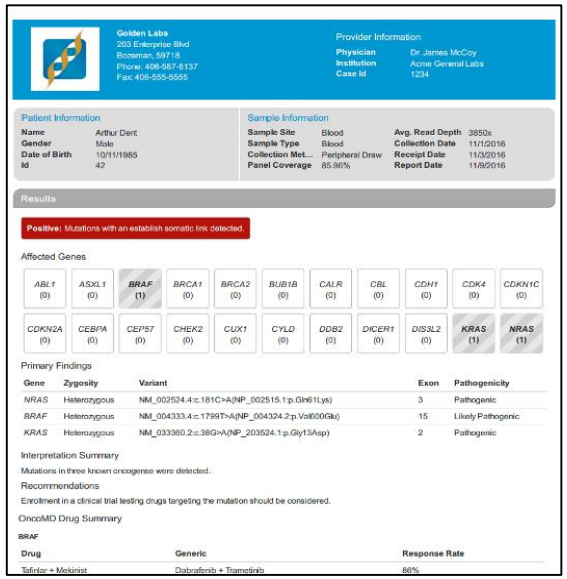
Gene	Exon	Variant	Zygosity	Pathogenicity
BRCA2	2	NM_000059.3:c.6513G>C	Heterozygous	Pathogenic

Interpretation Summary

A pathogenic variant has been detected in the BRCA2 gene. High risk of breast and ovarian cancer is associated with germline BRCA2 mutations.

Recommendations

Referral to an oncologist. Preventive or "prophylactic" mastectomy or removal of both breasts, has been found to reduce the risk of breast cancer in high-risk women by about 90% and should be considered.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Mutations in three known oncogenes were detected.

Recommendations

Enrollment in a clinical trial testing drugs targeting the mutation should be considered.

OncoMD Drug Summary

BRAF

Drug	Generic	Response Rate
Tafinlar + Mekinist	Dabrafenib + Trametinib	86%

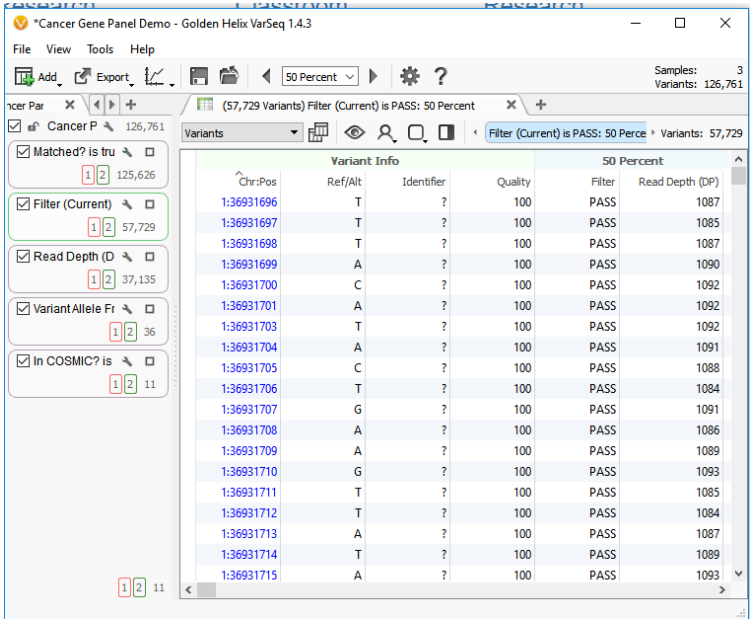
- **Generate Clinical Grade Reports**
  - ACMG Standard Germline Report
- **Reports are Fully Customizable**
  - Configurable Global Settings
  - Logo & Lab Information
  - Test Description / Disclaimers
- **Customizable Sample Inputs**
  - Patient Information & Test Results
- **Integrated with OMIM and OncoMD**
  - Functional Description Genes and Phenotypes
  - Drug Targeting Information
  - Ongoing Clinical Trials

Example Workflows

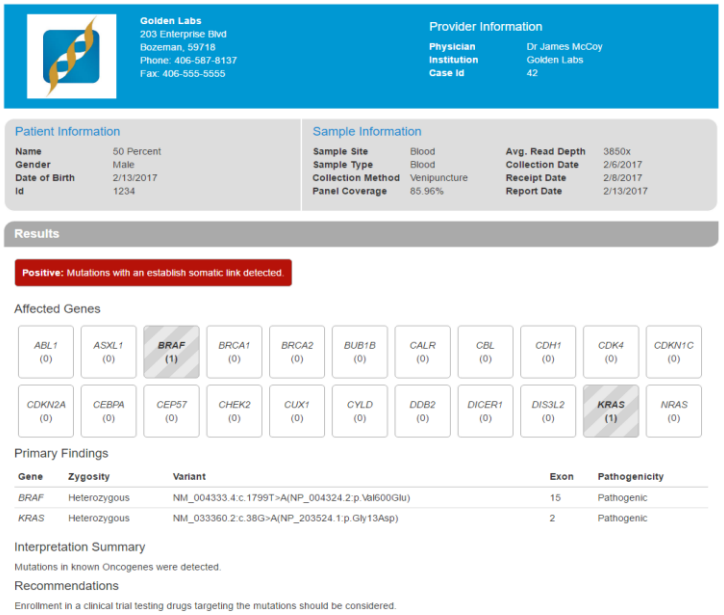
- **Illumina TruSight Myeloid Sequencing Panel**
  - Oncogenes & Tumor Suppressor Genes
    - Drug Targeting Information & Ongoing Clinical Trials
- **Exome Trio Workflow**
  - Casual Variants Associated with Phenotype
    - Incidental Findings
- **Illumina TruSight Cardio Sequencing Panel**
  - Cardiomyopathies, Arrhythmias, etc.
  - Integration with VSWarehouse

Sample Data for Cancer Gene Panels

- **Illumina TruSight Myeloid Sequencing Panel**
  - Comprehensive coverage of 54 genes designed to target exons of key tumor suppressor genes and frequently cited oncogenes.
  - BAM and VCF files for each replicate are available



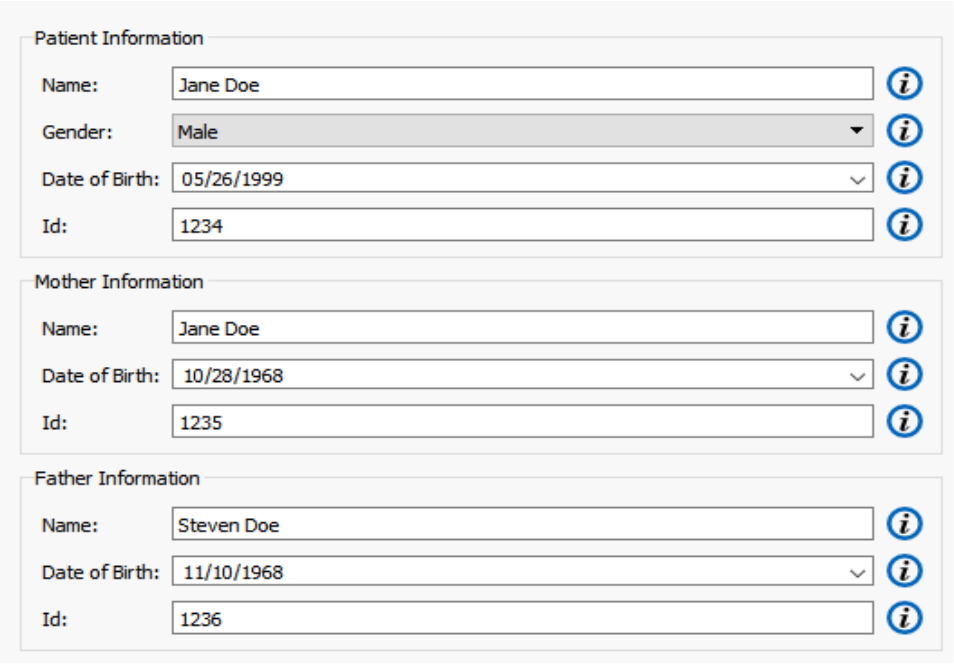
VSReports

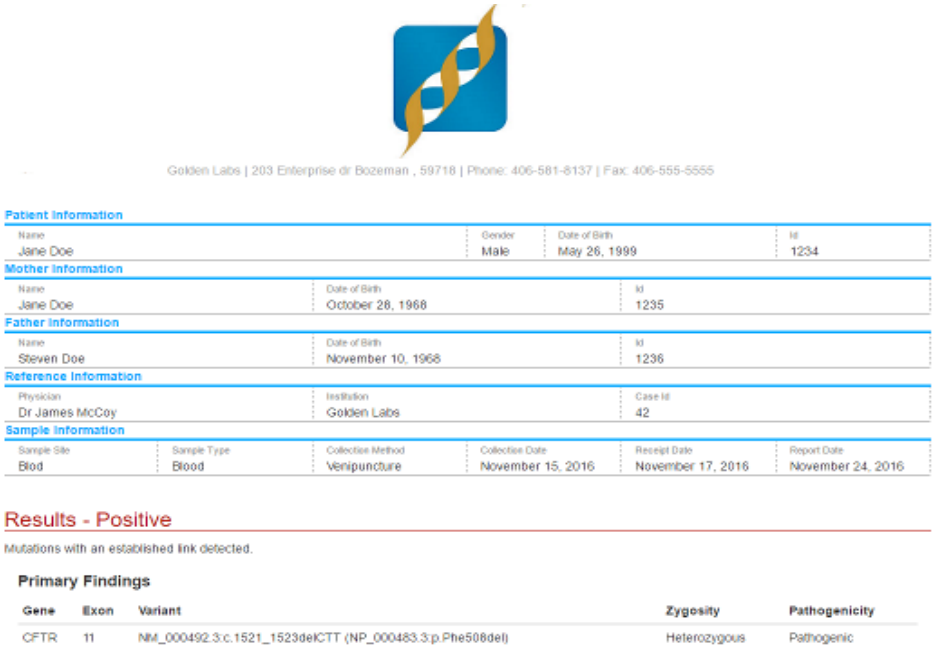


VarSeq Demonstration

Sample Data for Exome Trio Workflows

- **NA1278 17-year old Caucasian Women**
  - Admitted for Symptoms of Acute Gastroenteritis.
  - Serum testing revealed elevated pancreatic enzymes.
  - Genetic testing revealed Heterozygosity for CFTR gene mutation Δ F508
- **Incidental Findings**

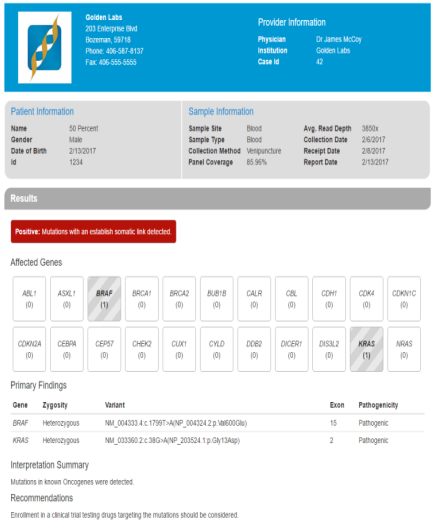




VarSeq Demonstration

Sample Data for Hereditary Gene Panels

- **TruSight Cardio Sequencing Kit from Illumina**
  - Comprehensive coverage of 174 genes with known associations to 17 inherited cardiac conditions (cardiomyopathies, arrhythmias, etc).



- **Have I ever seen this variant in my previous test samples?**

- **Have I put this variant into a clinical report for any previous samples?**

WAREHOUSE

- **Did I provide a pathogenicity assessment for this variant? Has that changed?**

Integration with VarSeq

- **VarSeq can "Export to Warehouse"**
  - Create a new Project
    - Current project template is used
  - Add Variants to Existing (data only)
- **Annotate with Warehouse Counts**
  - Allele / Genotype counts computed on warehouse variants (or other sources in template).
- **Variant Catalogs/ Annotation**
  - Annotate against variant catalog
- **Remote Reports**
  - Central versioning of report
  - Queryable database of all saved reports, with all data in form ready for EMR/LIMS integration

VarSeq Demonstration

Summary

- **VSReports & VarSeq**
  - Powerful Filtering & Annotation Engine Integrated with Clinical Reporting.
- **Clinical Report Customization**
  - Parameters, Layout, and Logo.
  - Golden Helix Offers Client Customizations.
- **Integration with VSWarehouse**
  - Track Reports, Variant Classifications.

GOLDEN HELIX

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New eBook Tomorrow!

NGS-Based Clinical Testing by Dr. Andreas Scherer

- Will be released tomorrow
- Chapter dedicated to clinical reporting

Special Offer

VarSeq Power Pack

- *First 3 customers only!*
- 2 seats for 15 months with access to VarSeq, VSReports, VSPipeline & VSWarehouse
- Access to OMIM & CADD included
- **$35,995!**
- Offer expires on March 31st, 2017 and is for new customers only
- Request a personal demo by emailing us **info@goldenhelix.com**

Questions or more info:

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