GOLDEN HELIX

Enabling Precision Medicine

An Exploration of Clinical Workflows in VarSeq

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CIOReview
20 most promising Biotech Technology Providers

pharma TECH OUTLOOK
Top 10 Analytics Solution Providers

Gartner
Hype Cycle for Life sciences

Golden Helix – Who We Are

Golden Helix is a global bioinformatics company founded in 1998.

GlaxoSmithKline

VarSEQ®

- Variant Calling
- Filtering and Annotation
- Clinical Reports
- CNV Analysis
- Pipeline: Run Workflows

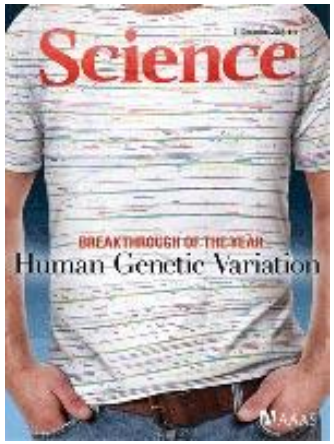
VARIANT WAREHOUSE

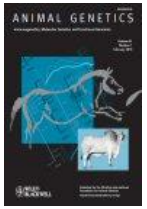
- Variant Warehouse
- Centralized Annotations
- Hosted Reports
- Sharing and Integration

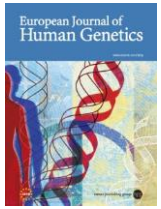
SNP & VARIATION SUITE

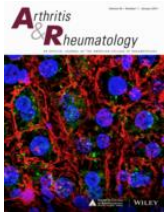
- GWAS
- Genomic Prediction
- Large-N-Population Studies
- RNA-Seq
- Large-N CNV-Analysis

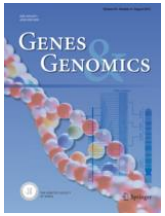
Cited in over 1100 peer-reviewed publications











GOLDEN HELIX
Enabling Precision Medicine

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

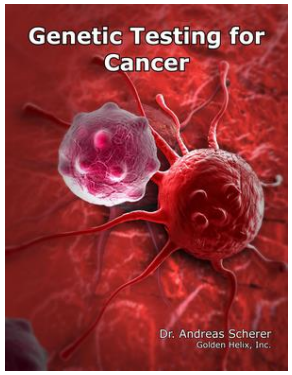
FDA

GOLDEN HELIX
Enabling Precision Medicine

Golden Helix – Who We Are

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

- REPUTATION
- TRUST
- EXPERIENCE

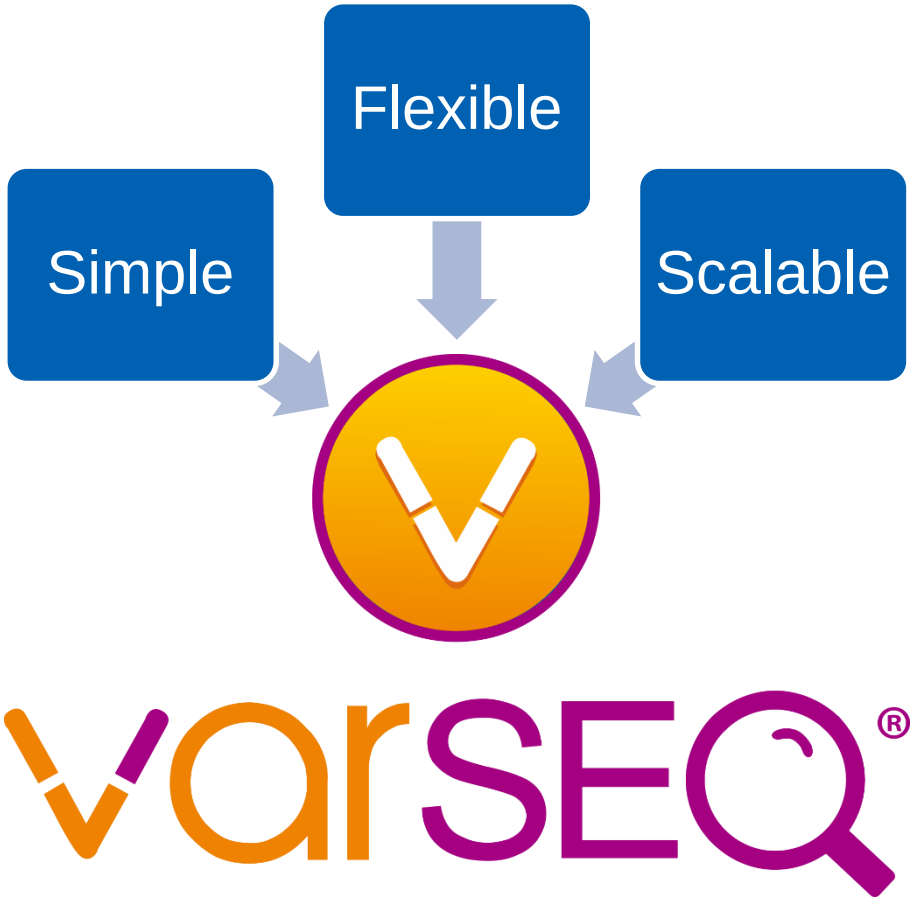


- INDUSTRY FOCUS
- THOUGHT LEADERSHIP
- COMMUNITY

- TRAINING
- SUPPORT
- RESPONSIVENESS

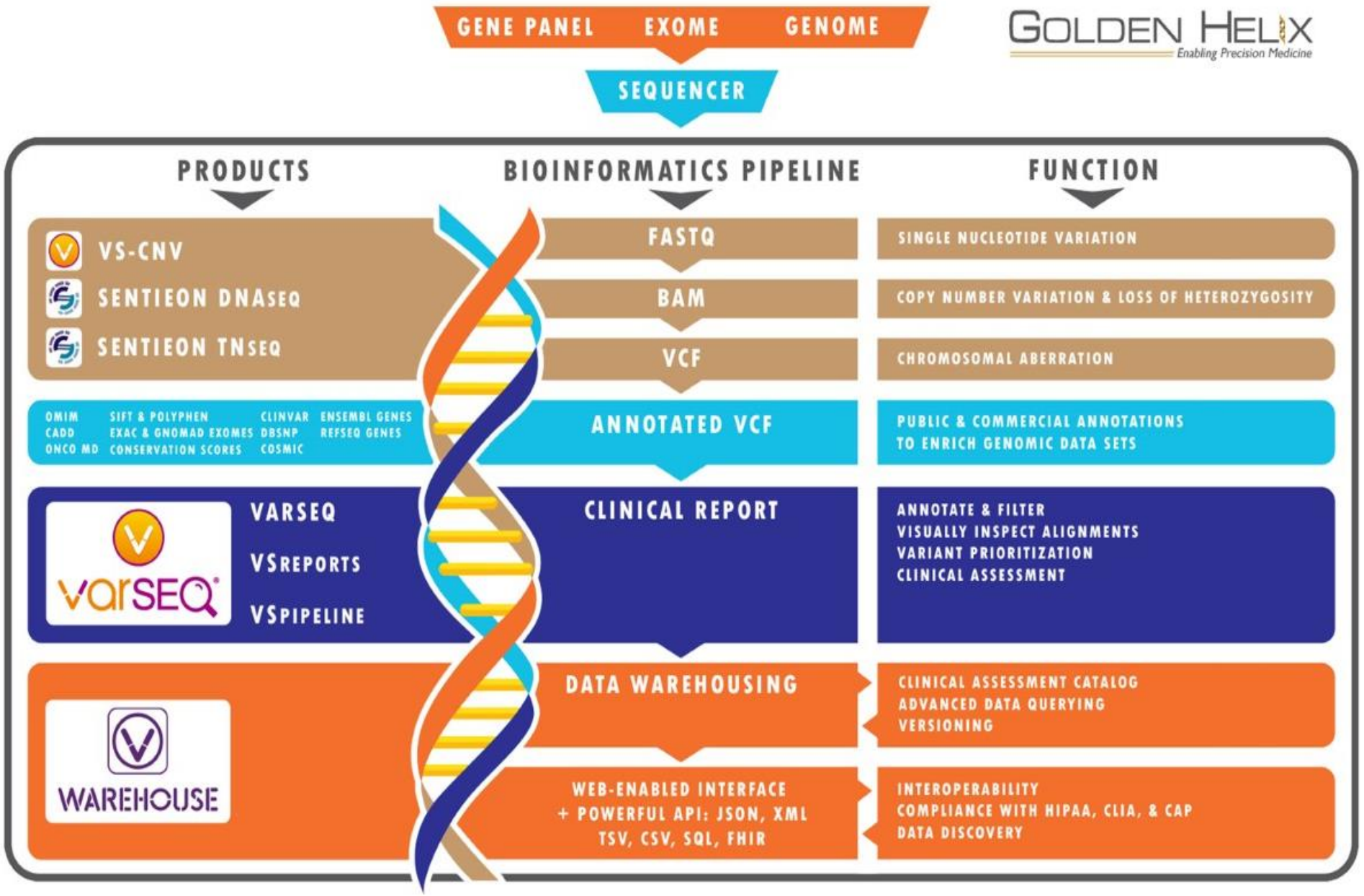
- TRANSPARENCY
- INNOVATION and SPEED
- CUSTOMIZATIONS

VarSeq Suite

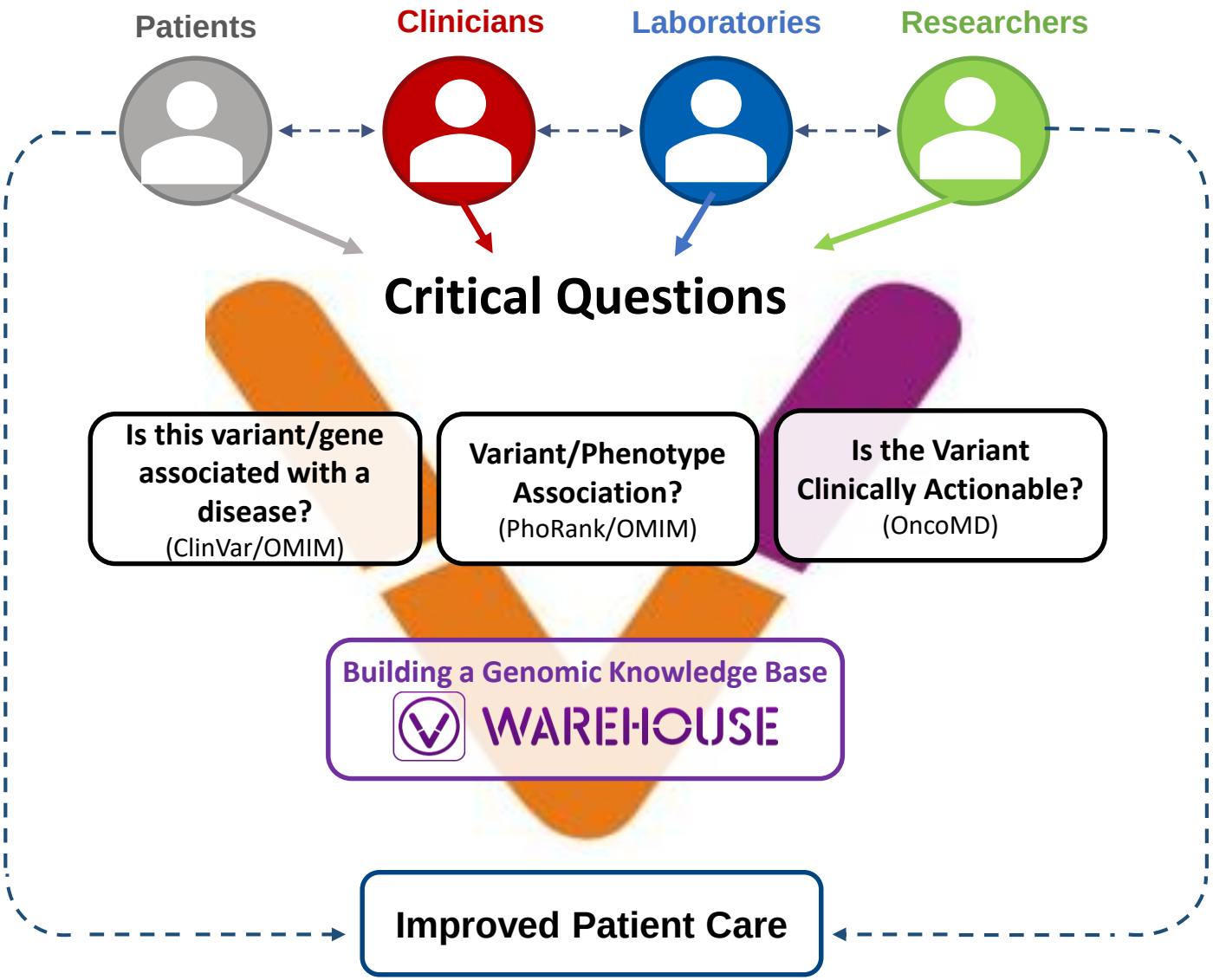


- **Variant annotation, filtering, and interpretation**
- **Powerful GUI with rich visualizations**
- **Repeatable workflows & command-line interfaces**

VarSeq Complete Stack



VarSeq Clinical Workflow



TruSight Cancer Gene Panels

- **TruSight Cancer Sequencing Kit from Illumina**
  - 6 samples; 1 with Lung Cancer Phenotype
  - Key mutations in Tumor Suppressor Genes or Known Oncogenes?

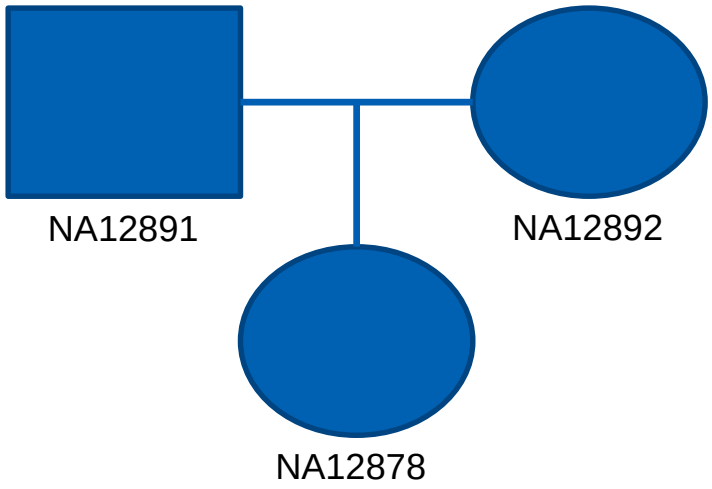
- **Drugs Target Protein Products of Oncogenes**
  - Epidermal Growth Factor (EGFR)
  - Anaplastic lymphoma kinase (ALK).
    - Crizotinib & Ceritinib – inhibitors of oncogenesis.

- **Does this sample possess key variants in Oncogenes?**
  - If so, is it pathogenic?
  - Drug targeting information, ongoing clinical trials?
    - Create a clinical report.

VarSeq Demonstration

Trio Exome Analysis

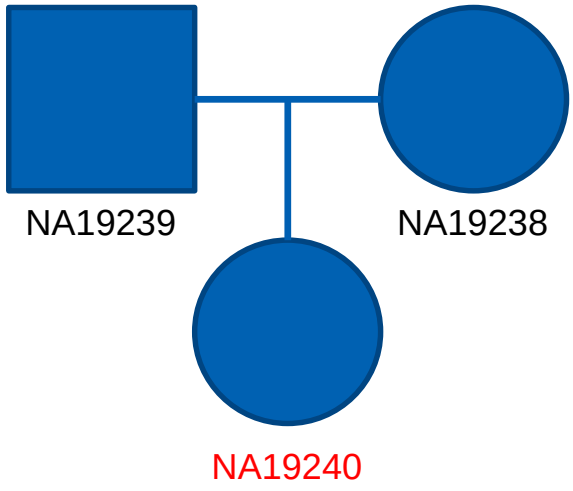
- **From International HapMap Project**
  - BAM file from 1000 Genomes Phase 3 Illumina Exome Alignment
  - Used Sentieon tools for variant calling.
  - Mutations in the CTFR gene (cystic fibrosis) and BRCA1 gene (breast cancer)



VarSeq Demonstration

Single Exome Analysis

- **NA19240 from International HapMap Project – Yoruba in Ibadan, Nigeria**
  - BAM file from 1000 Genomes Phase 3 Illumina Exome Alignment
  - Used Sentieon tools for variant calling.
  - Has a mutation in the SMAD4 gene which is associated with Myrhe Syndrome.



VarSeq Demonstration

Questions or more info:

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