Introducing
SNP & Variation Suite 8

March 11, 2014

Gabe Rudy
VP Product Development

GOLDEN HELIX
Accelerating the Quest for Significance™

Introduction

- SVS 7 has been adopted by hundreds of client organizations worldwide.
- GenomeBrowse has 2,500 registered users.

Andreas Scherer, PhD, President & CEO

Introduction

Researchers

- Human genome
- Plant DNA
- Animal DNA

Clinicians

- Also use genomic information to:
- Diagnose diseases
- Identify best treatment options for patients

Students

- Currently need to be a “computer science” whiz before they can begin analyzing data
- SVS reduces the initial hurdle to enter the field

Introduction

- Sequencing technology becoming cheaper
- Scalability is key
- Sample sizes are increasing
- More data per sample
- **Improvement of:**
  - Data management
  - Data import and manipulation capabilities

Introduction

Company maturity:

- Service engagements
- Deeply ingrained in the community
- Hosting webcasts
- Website becomes knowledge hub

our 2 snps...

A BLOG BY GOLDEN HELIX

Home Authors @gabeinformatics

← First Golden Helix Research Competition a Great Success!

The Next Chapter of Golden Helix: Our Launch of SVS 8

Posted on March 10, 2014 by goldenadmin

Today is a big day for us. Today we are launching [JV1] a major release of our flagship product, SNP & Variation Suite (SVS), to the general public. SVS 8 is a substantial improvement over the previous release in a number of dimensions (see detailed discussion on our What's New page).

We've come a long way.

Over five years ago, in November 2008, we introduced SVS 7 as a powerful tool to conduct next-generation sequencing and GWAS studies. Since then, we have been adopted by hundreds of client organization worldwide. SVS is being used by leading research organizations in the US, Canada, Latin America, Asia, Australia, Africa, and Europe.

About one and a half years ago, we launched the first version of our free, standalone genome browser. This tool was designed to help researchers to view large sequencing files alongside public annotation databases in a fluid and intuitive way. Over 2,500 researchers in our field are using GenomeBrowse today.

As customers have seen the intuitive way GenomeBrowse allows them to navigate through extensive databases and large amount of data, we have been asked to bring the same visualization look and feel to SVS (which is the biggest change for SVS 8). This becomes increasingly important as SVS is adopted outside of our traditional audience.

Our primary users are researchers who analyze human, plant, or animal DNA to study diseases and traits. Our software is also used in clinical settings where scientists are working hand-in-hand with their clinical counterparts to use genomic information to diagnose diseases and identify best treatment options for their patients. In addition, we see a third group of users becoming increasingly important: undergrads and graduate students who are learning how to conduct analytics work. Here, the ease-of-use of our platform reduces the initial hurdle to enter the field. It eliminates the need for a student to become a biostatistics whiz before they can begin analyzing data and focusing on the science behind it.

Another major topic that we addressed in this release is scalability. As sequencing technology has

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About

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- Add-on scripts & data repository (12)
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- Big picture (21)
- Bioinformatic support (9)
- Clinical genetics (6)
- Customer success (18)
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- News, events, & announcements (44)
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Questions during the presentation

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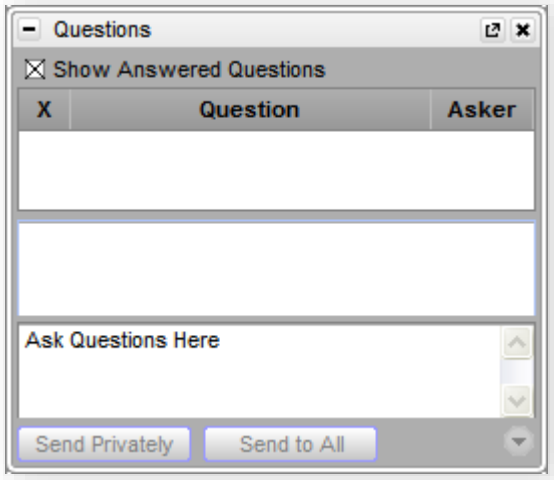
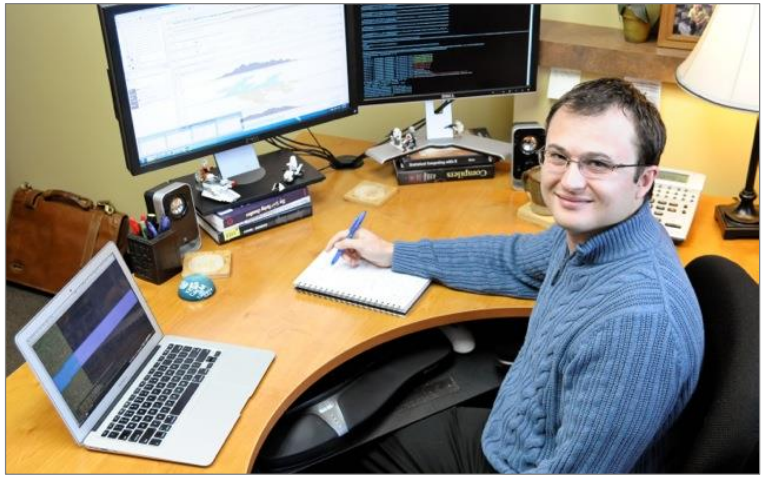


Agenda

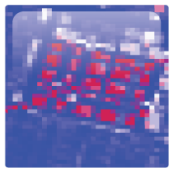
1. History
2. New Annotation Infrastructure
3. GenomeBrowse Grows in Its Utility
4. What's New in GWAS Analysis
5. Conclusion

Background

- **Golden Helix**
  - Founded in 1998
  - Genetic association software
  - Analytic services
  - Hundreds of users worldwide
  - Over 800 customer citations in scientific journals
- **Products I Build with My Team**
  - **SNP & Variation Suite (SVS)**
    - SNP, CNV, NGS tertiary analysis
    - Import and deal with all flavors of upstream data
  - **GenomeBrowse**
    - Visualization of everything with genomic coordinates. All standardized file formats.
  - **RNA-Seq Pipeline**
    - Expression profiling bioinformatics



Historical Context



HT 6

Oct 2007

SVS 7

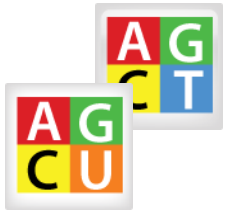
Feb 2009

7.3

Jul 2010

7.5

Jul 2011



7.7

Nov 2012

MLM

7.7.8

Aug 2013

SVS 8

May 2011

Design

Mar 2012

Beta

Sep 2012

1.0

Mar 2013

1.1

VCF

Integrated Product Solution

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Annotations

- Annotations are the cornerstone of DNA analysis and interpretation
- **Our rebuilt infrastructure results in:**
  - New **file format** that provides compressed files with richer usage and indexed search
  - New **Convert** wizard
  - Update Annotation and Filtering **workflows** to utilize these sources
  - Redesigned **data repository** with the future in mind

File Format

- Amazing in-place compression with column data storage techniques
- Scales to hundreds of millions of records without slowing down
- Integrated documentation
- Indexed field searching

@gabeinformatics

AN "OUR 2 SNPs..." BLOG BY GOLDEN HELIX

Home Authors @gabeinformatics

← CALL FOR ABSTRACTS: Dell Ultrabook Laptop and SVS Giveaway

Back to Basics: Importing/Exporting Data in Imputation Program Data Formats with SVS →

All I Want for Christmas Is a New File Format for Genomics

Posted on December 16, 2013 by Gabe Rudy

'Tis the season of quiet, productive hours. I've been spending a lot of mine thinking about file formats. Actually I've been spending mine implementing a new one, but more on that later.

File formats are amazingly important in big data science. In genomics, it is hard not to be awed by how successful the BAM file format is.

I thought one of the most tweetable moments at ASHG 2013 was when Jeffrey Reid from BCM Human Genome Sequencing Center (HGSC) talked about how they offloaded to the cloud (via DNAnexus) 2.4 millllion hours of compute time to perform the alignment and variant calling on ~4k genomes and ~12k exomes.

Heng LI thinking up another algorithm...

In the process, they produced roughly half a petabyte of BAM files (well mostly BAM files, VCFs are an order of magnitude smaller, but part of the output mix).

I'd speculate that Heng LI's binary file format for storing alignments of short reads to a reference genome is responsible for more bytes of data being stored on the cloud (and maybe in general) than any other file format in the mere 4 years since it was invented.

But really, the genius of the format was not in the clever and extensible encoding of the output of alignment algorithms (the CIGAR string and key-value pair "tag" field have held up remarkably well through years of innovation and dozens of tools), but in the one-to-one relationship it shared with its text-based counterpart, the SAM file.

Wanted: Grey Beard Unix Hackers for Big Data Science

Text files have been, and will continue to be, the dominant medium of data scientists – and for good reasons. Even a basic familiarity with the Unix toolset allows for an immense amount of data management and processing tasks to take place. And to take place efficiently. I dare you to write a program that is faster at searching text files than grep.

In fact, any bioinformatic pipeline worth its salt is probably heavily utilizing grep, awk, cut, sed, sort, xargs and, yes, the rusty edged Swiss army knife that is perl. We've worked on a couple here at

Search

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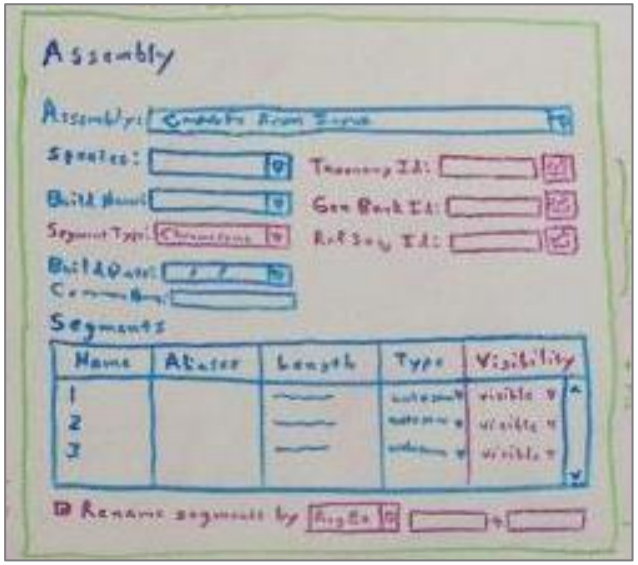
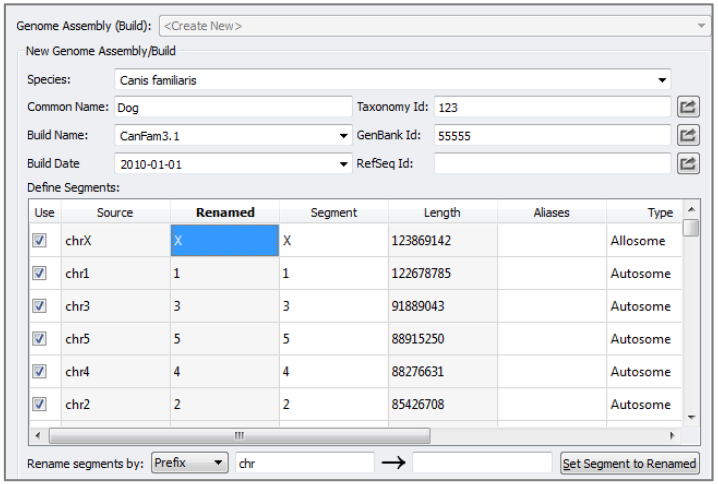
- Plant & animal (6)
- Technology review (25)

Convert Wizard

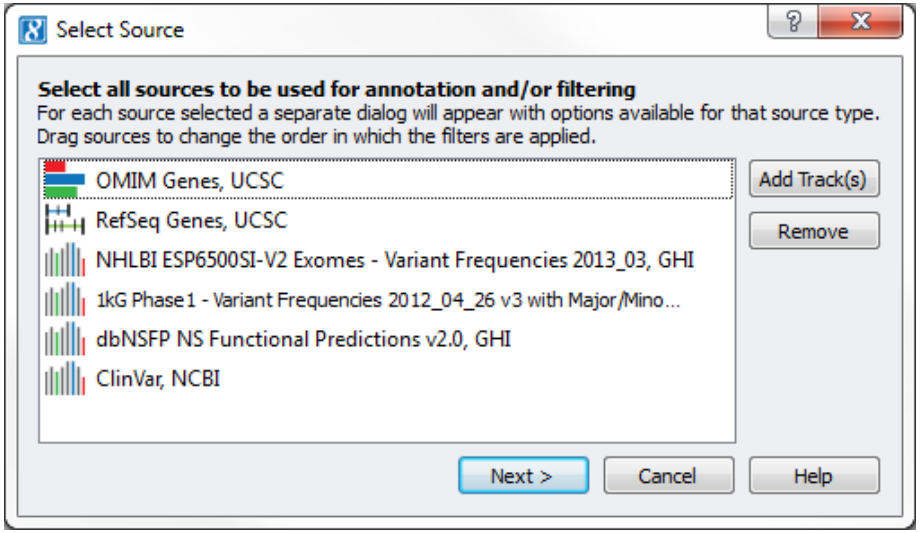
- Supports new file formats – every type of file we have seen, we can convert
- Preview your data as you go
- Document your data source in the wizard or afterwards
- Curate your own genomes
- Easy to share your results – just one file

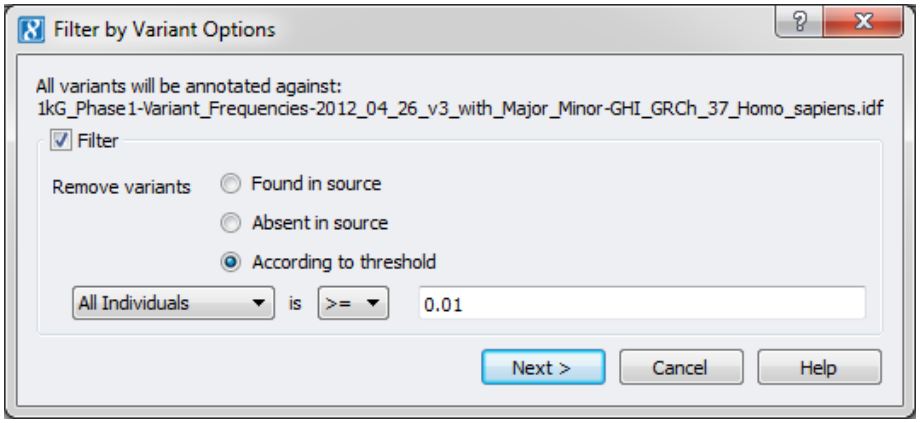




Annotation Workflow

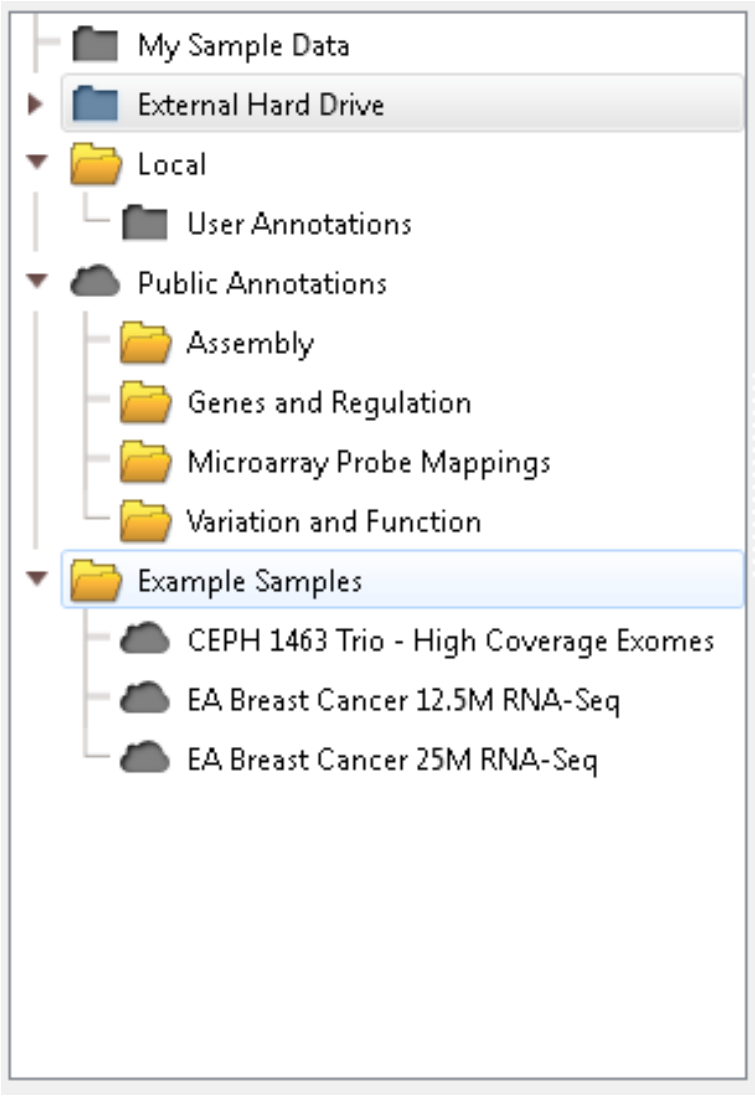
- Pick all your annotations up front, be guided through choices on each one
- Annotate smaller datasets over the network
- Chain your filters





Data Repository

- Have hierarchy and most important use versioning
- We never delete a previously published data source. Access any version at any time.
- Fast and easy streaming, or download using our new downloader
- Documentation built in
- Historical repository converted to TSF, but your existing files will continue to work.



[Demo]

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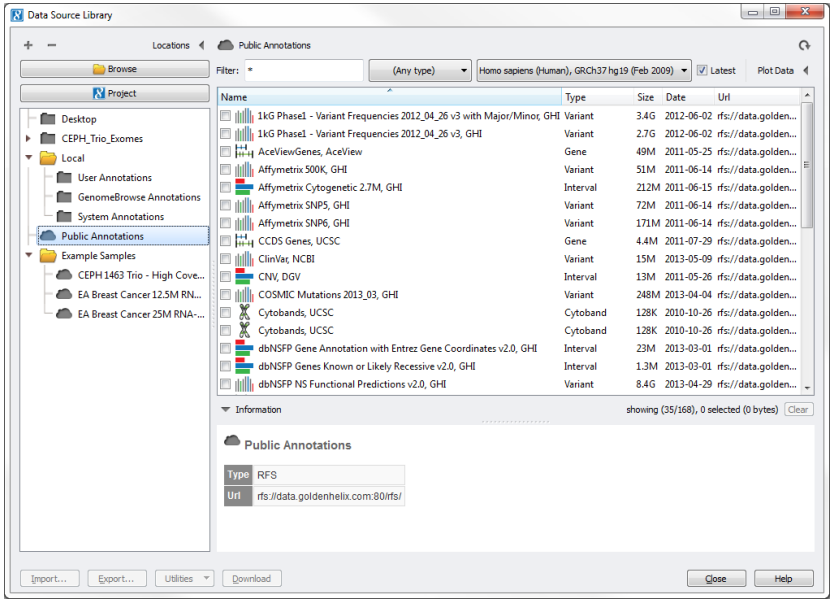
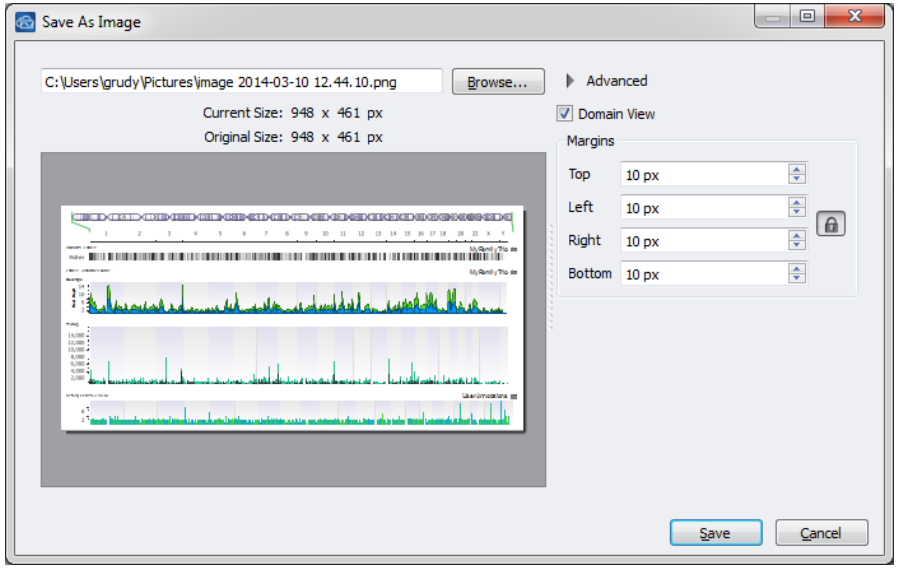
Data Source Library

- Re-imagined GenomeBrowse's data management interface
  - Provides integrated preview of documentation
  - Easily add permanent folders to be in your library (with ability to be recursive)
  - Browse any folder to quickly find a plottable source



Save as Image

- Have had people tell us that they love GenomeBrowse as its renderings are better than publication quality
- Needed a flexible way to export that beautiful rendering
- We didn't want to cut corners, and the use cases are quite complex
- Have rewritten half of GenomeBrowse to be in a flexible architecture for all the plot rendering to be laid out and rendered to devices other than the screen.



- For this release, we have a great start, but have more to come

[Demo]

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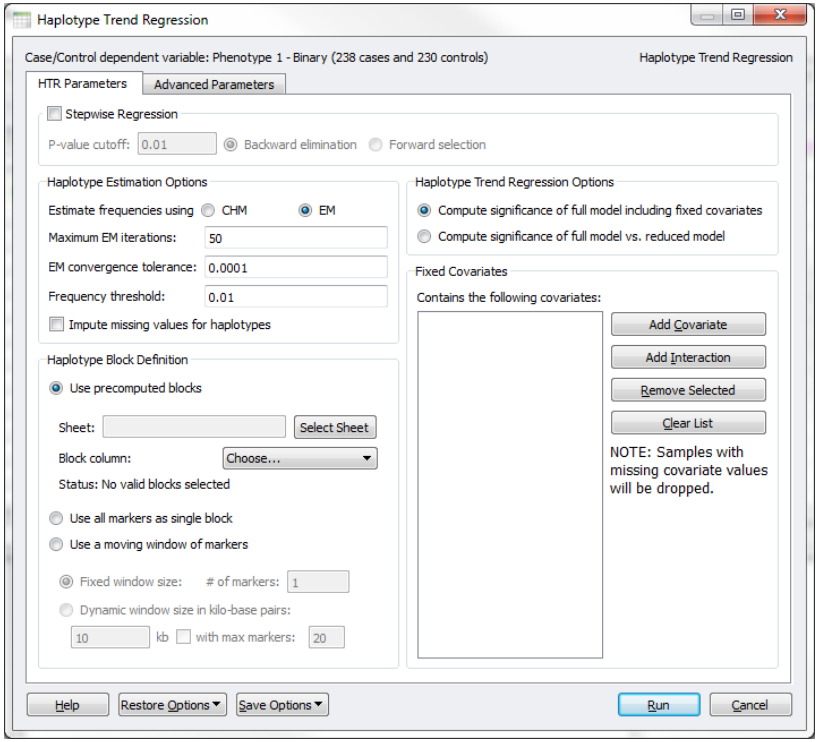
Haplotype Trend Regression

- Haplotype Trend Regression one of our most requested features from our HelixTree line that wasn't supported in SVS 7
- Now SVS 8 has HTR with more:
  - Marker blocks
  - Covariates
  - Can use binary or continue dependent variable



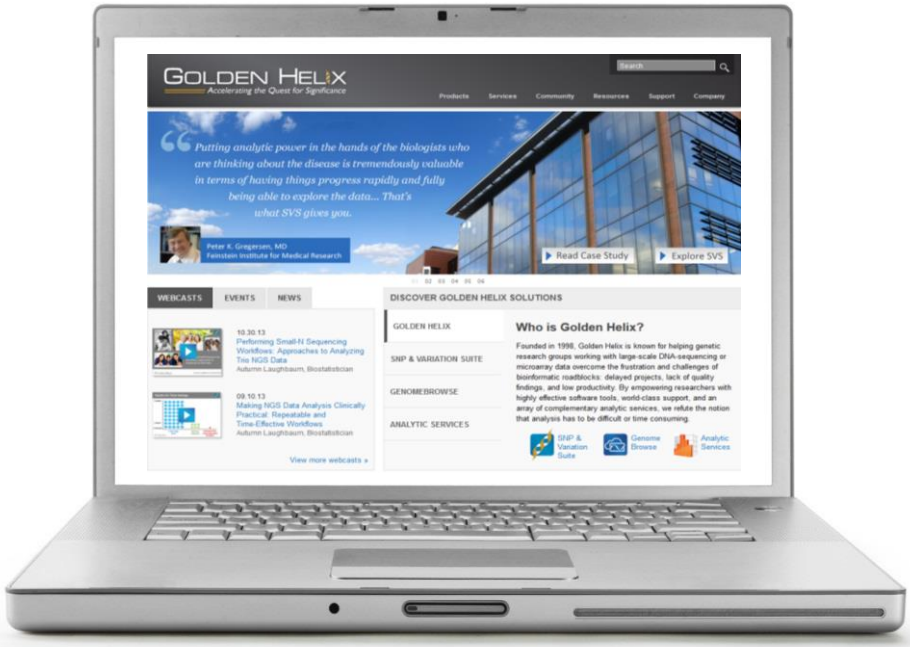
[Demo]

Other New Features

- ***Fst* by Marker**
- **DNA-Seq Additions**
  - Classify by Inheritance Pattern
  - Calculate Alt Read Ratio
  - Score Variants by Dominant Model
  - Filter based on VCF Quality Metrics

Updates are always free for customers

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



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