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
Company maturity:

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

Use the Questions pane in your GoToWebinar window
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
Integrated Product Solution
Agenda

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

1. History

2. New Annotation Infrastructure

3. **GenomeBrowse Grows in Its Utility**

4. What’s New in GWAS Analysis

5. Conclusion
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
- 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]
Agenda

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

Use the Questions pane in your GoToWebinar window.