

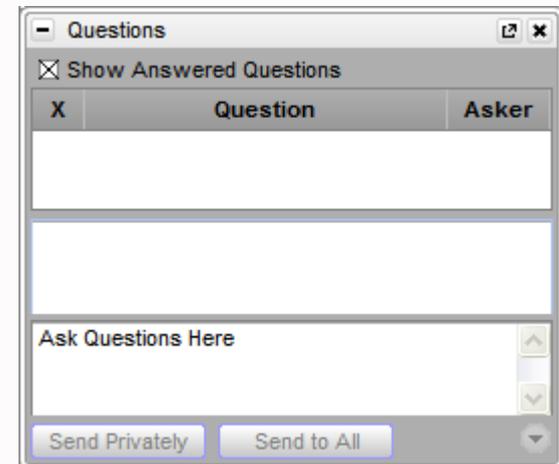
Performing Small-N Sequencing Workflows: Approaches to Analyzing Trio NGS Data



Questions during the presentation



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Today's Agenda



1 Overview

- Why perform sequence analysis using small families?
- Workflow Approach: Data Quality Preparation, Initial Investigation, Analysis

2 SNP & Variation Suite (SVS)

- Python Interface

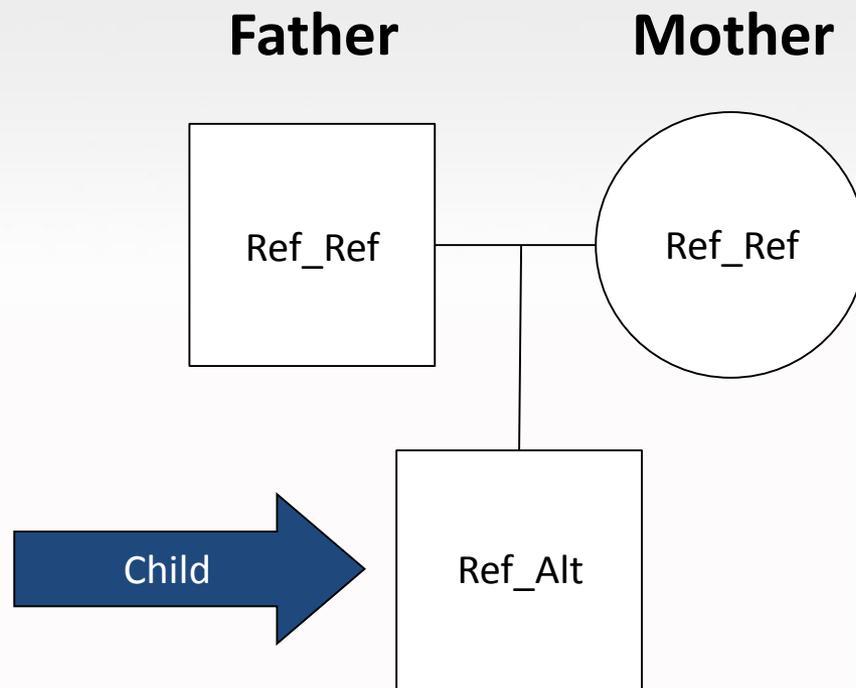
3 SVS Demo

4 Conclusion

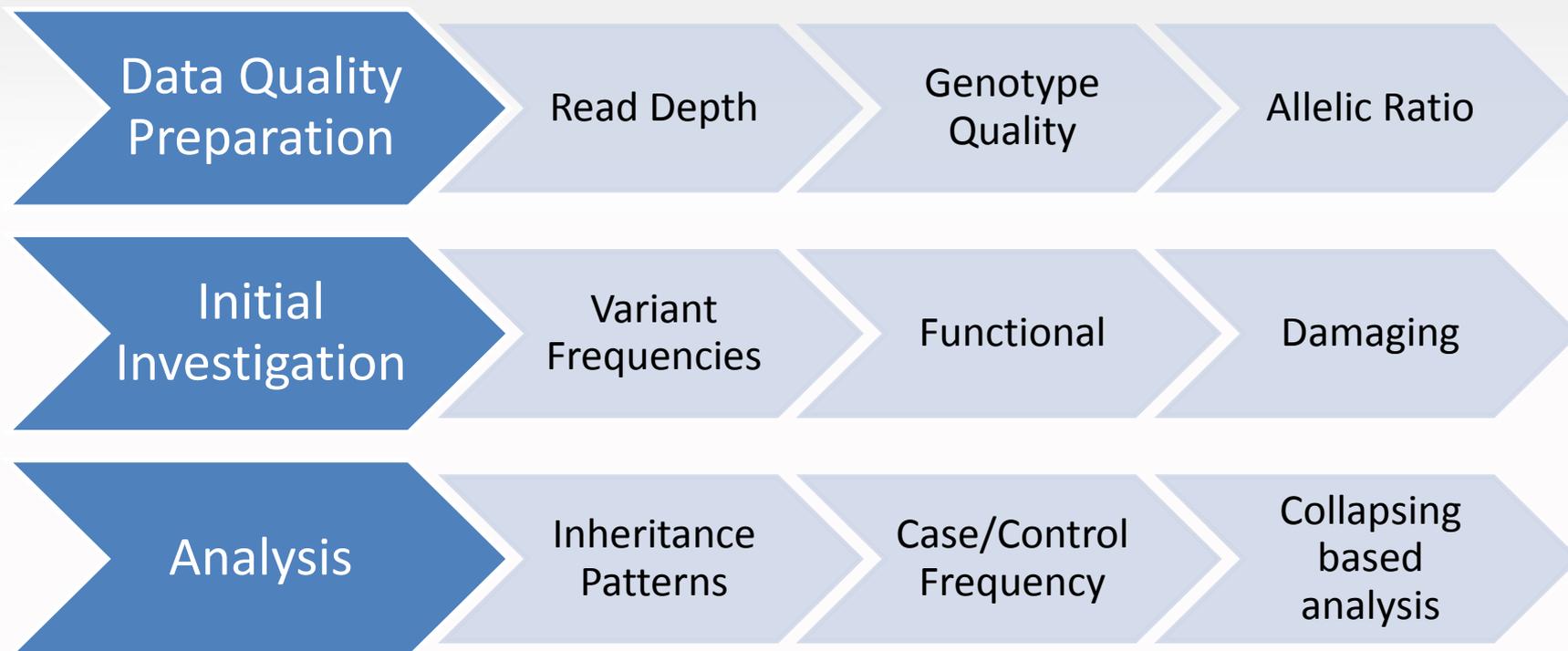


Overview

Why perform seq analysis using small families?



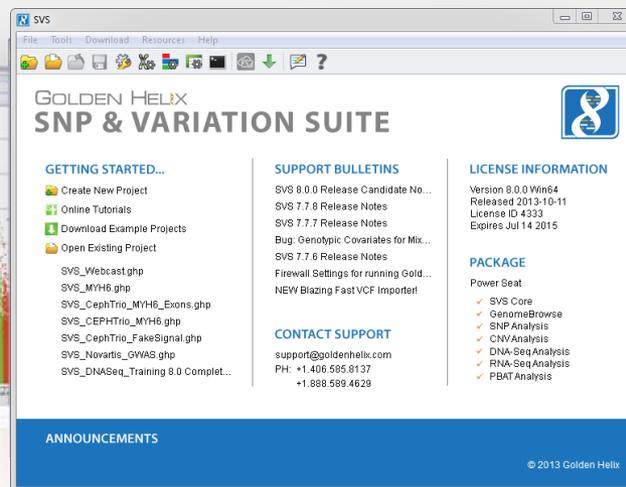
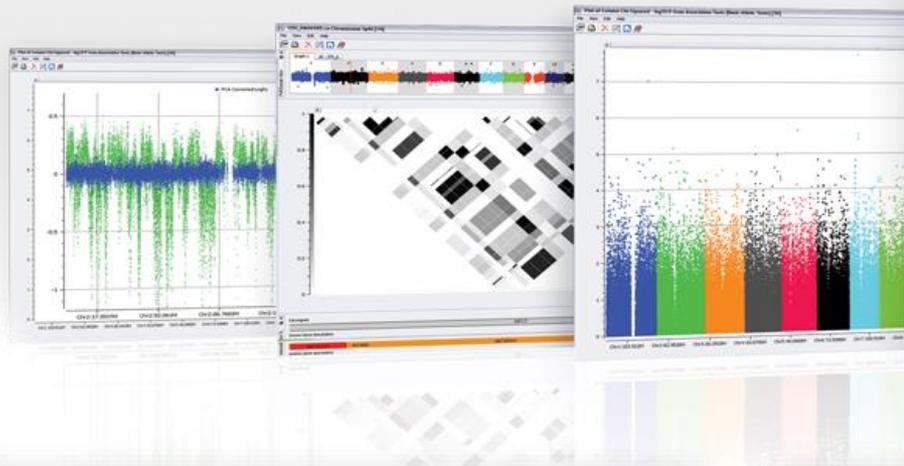
The 3-Part Workflow Approach





SNP & Variation Suite (SVS)

SNP & Variation Suite (SVS)



Core Features

- Powerful Data Management
- Rich Visualizations
- Robust Statistics
- Flexible
- Easy-to-use

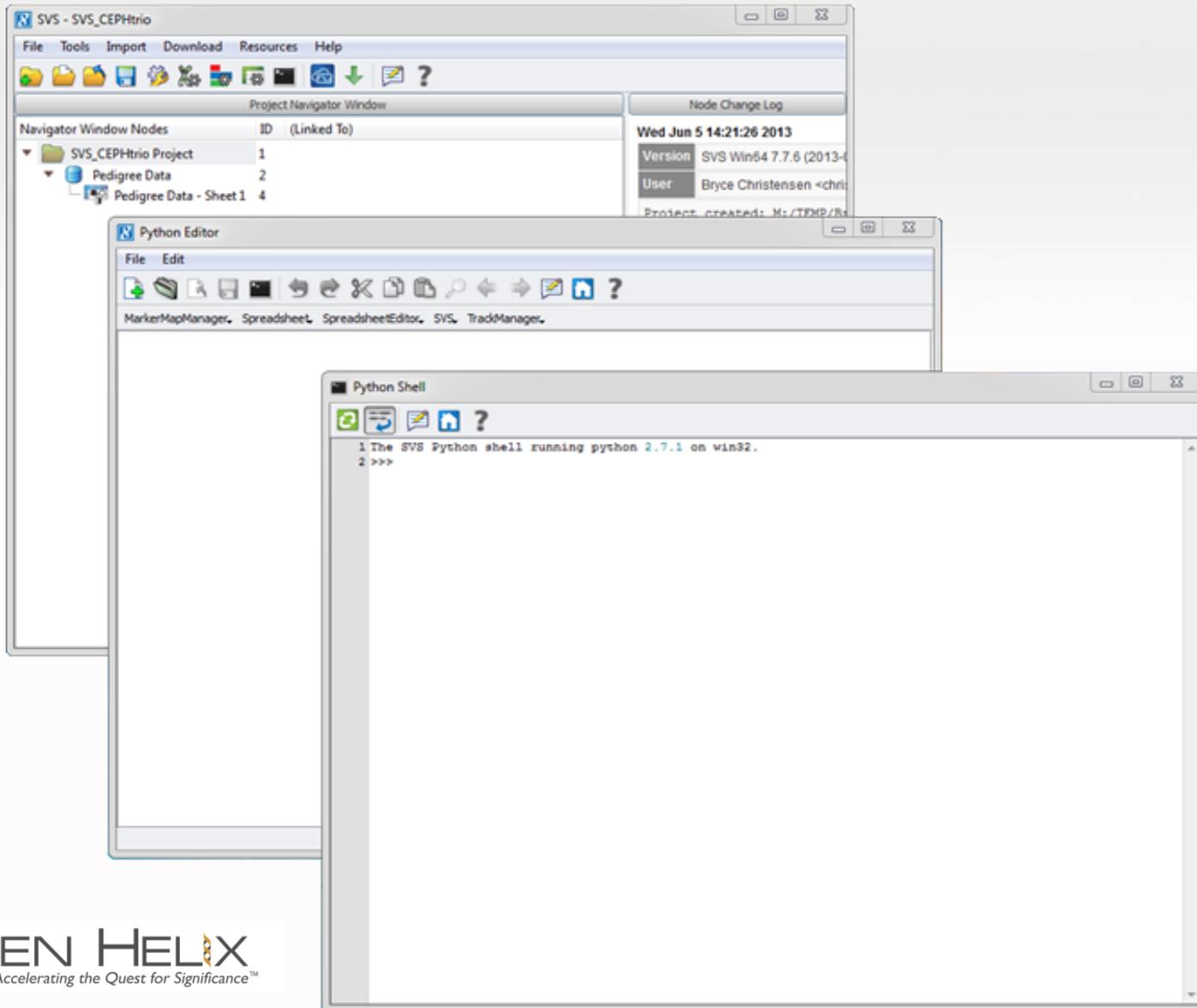
Applications

- Genotype Analysis
- DNA sequence analysis
- CNV Analysis
- RNA-seq differential expression
- Family Based Association



Python Interface

Python Interface



SVS Add-On Scripts Repository



SVS Add-On Scripts Repository

goldenhelix.com/SNP_Variation/scripts/index.html

Apps Tasks Golden Helix R&D | ... GHI Mail FogBugz Golden Helix, Inc Technical Support B... Latest Documentation SVS Manual - Stable Tutorials: SNP and V... SVS Add-On Scripts ...

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- SVS Manual
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OUR 2 SNPs...[®] GHI Blog

"Precision Medicine": Moving Next-Generation Sequencing into the Clinic

Add-On Scripts Repository for SVS

Here you will find a collection of [Python scripts](#) submitted by Golden Helix developers and our customers. All scripts are provided for no additional cost. So feel free to download, use, and even enhance!

The following scripts are for **SVS 7.4+**
For scripts compatible with older versions, please visit the [Scripts Repository for SVS 7.0-7.3](#).

Share your scripts with the Golden Helix Community

If you have written any scripts and would like to share them with other SVS 7 users, we encourage you to email a *.txt or *.py file to community@goldenhelix.com with any accompanying documentation or special instructions. Once we test your script and check its validity, we'll post it on this page for others to download.

What is Python?

Python is a clear and powerful object-oriented programming language, comparable to Perl, Ruby, Scheme, or Java. Integrating Python into SVS 7 provides full programmatic access to many of the software's features enabling the augmentation of existing tools, creating entirely new ones, automation of work flows, integration with other programs and more.

Python Learning Resources

- » SVS 7 Scripting Reference
- » Python.org
- » Beginners Guide to Python

Keep informed on new scripts by subscribing to the [technical support bulletins feed](#) »

Date Modified	Category	Script	Author	Download
8/26/2013	Filter	Subset by Chromosome This script scans genetic marker mapped columns and creates a subset spreadsheet for each unique chromosome with active data in the spreadsheet. More info »	Autumn Laughbaum Golden Helix	
8/26/2013	Filter	Inactivate Duplicate Row Values This script scans a selected column in a spreadsheet and inactivates rows based on user prompts by either inactivating all copies of the duplicate values or keeping the first occurrence and inactivating all subsequent duplicates. Row values need to match exactly, including case, to be consider duplicates. More info »	Christophe Lambert Golden Helix	



SVS Demo

Exome Sequencing









[Poll]



Conclusion



Questions?

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