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

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Autumn Laughbaum, Biostatistician

Questions during the presentation

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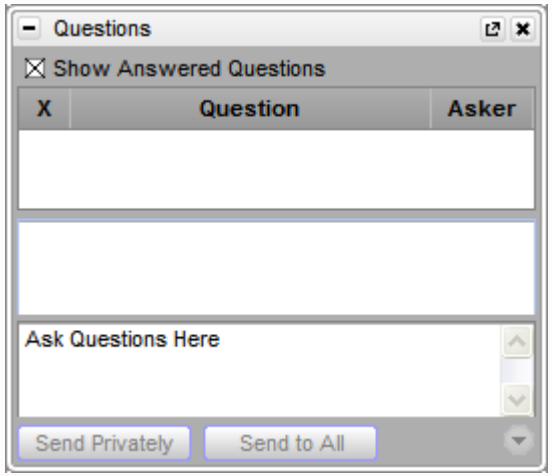

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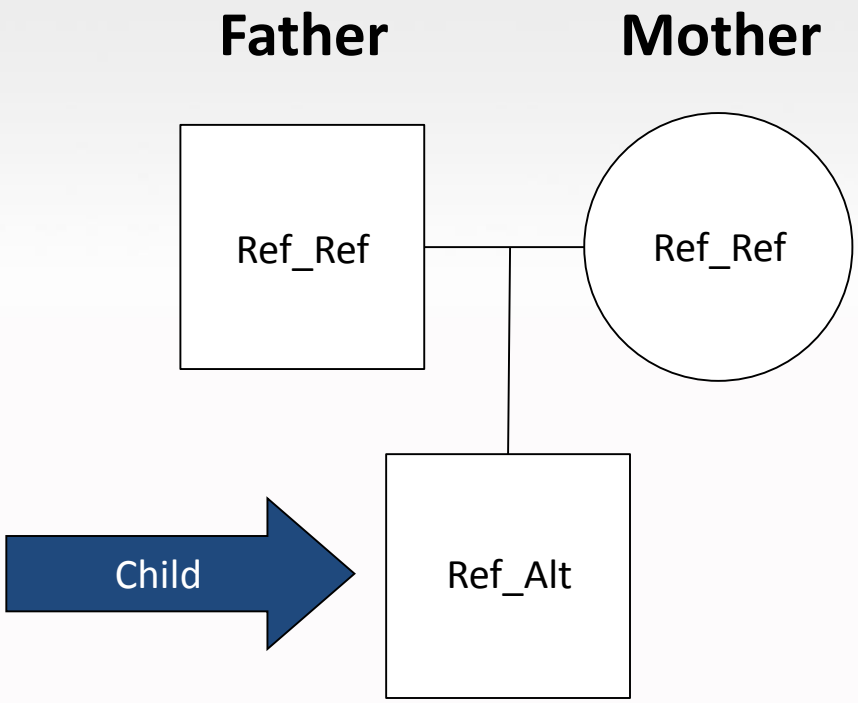
Today's Agenda

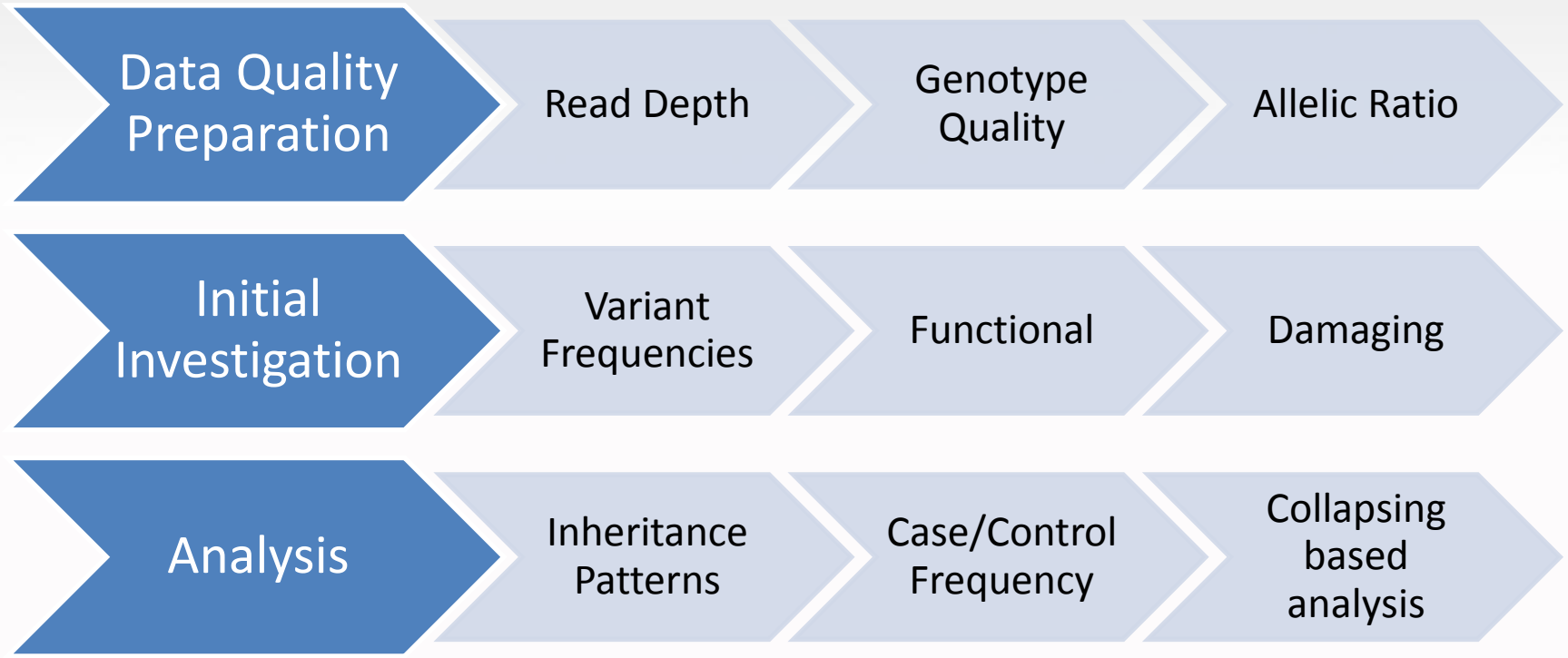
1. Overview
   - Why perform sequence analysis using small families?
   - Workflow Approach: Data Quality Preparation, Initial Investigation, Analysis
2. SNP & Variation Suite (SVS)
   - Python Interface
3. SVS Demo
4. Conclusion

Overview

Why perform seq analysis using small families?



The 3-Part Workflow Approach



SNP & Variation Suite (SVS)

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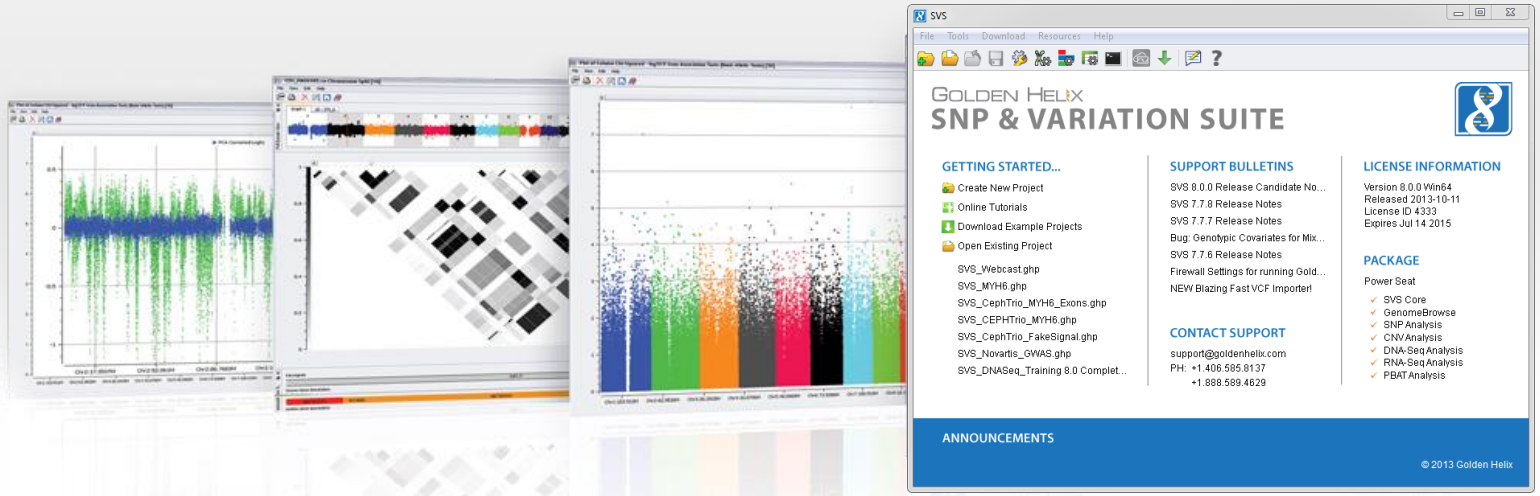
SNP & Variation Suite (SVS)



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

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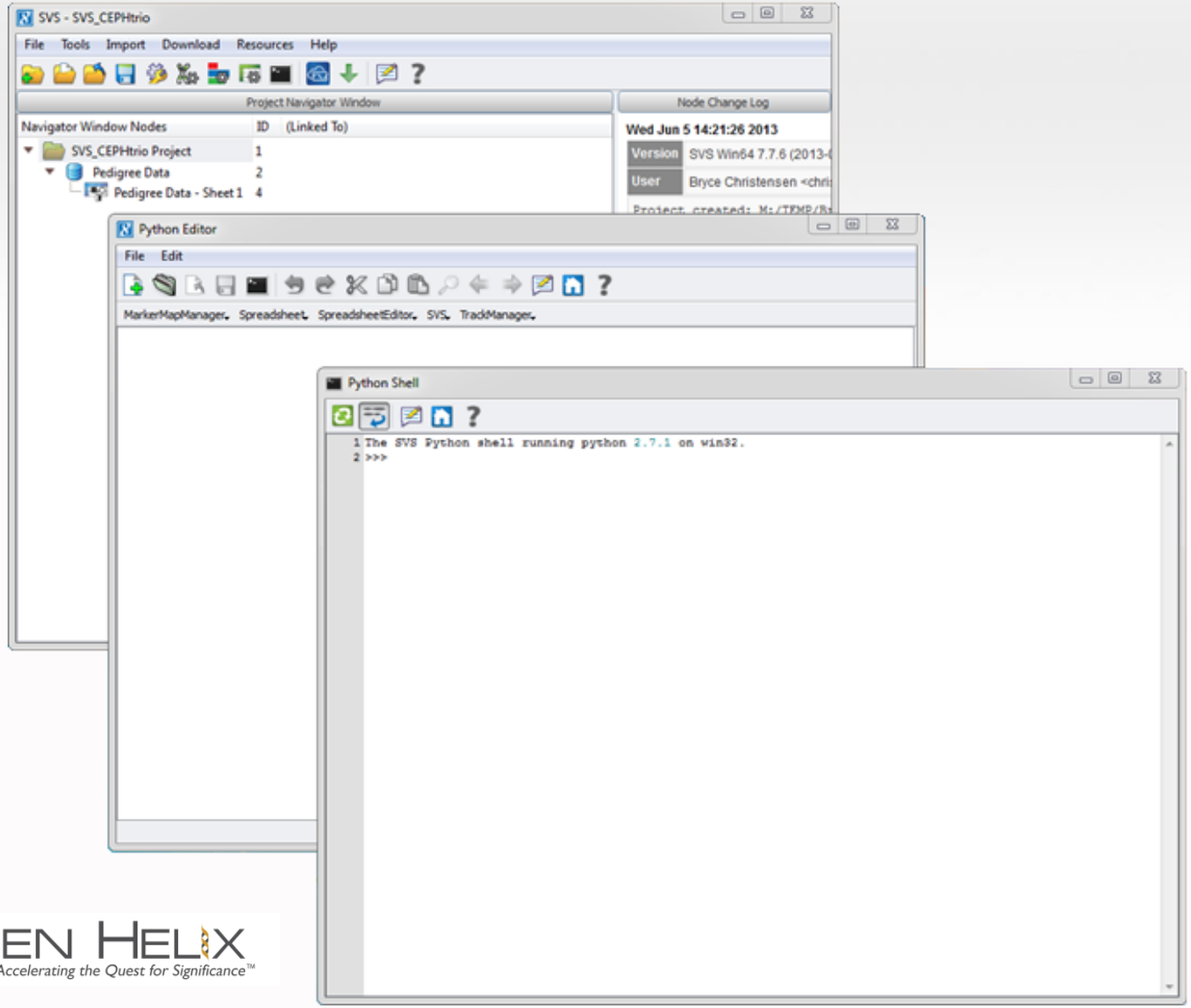
Python Interface

Python Interface



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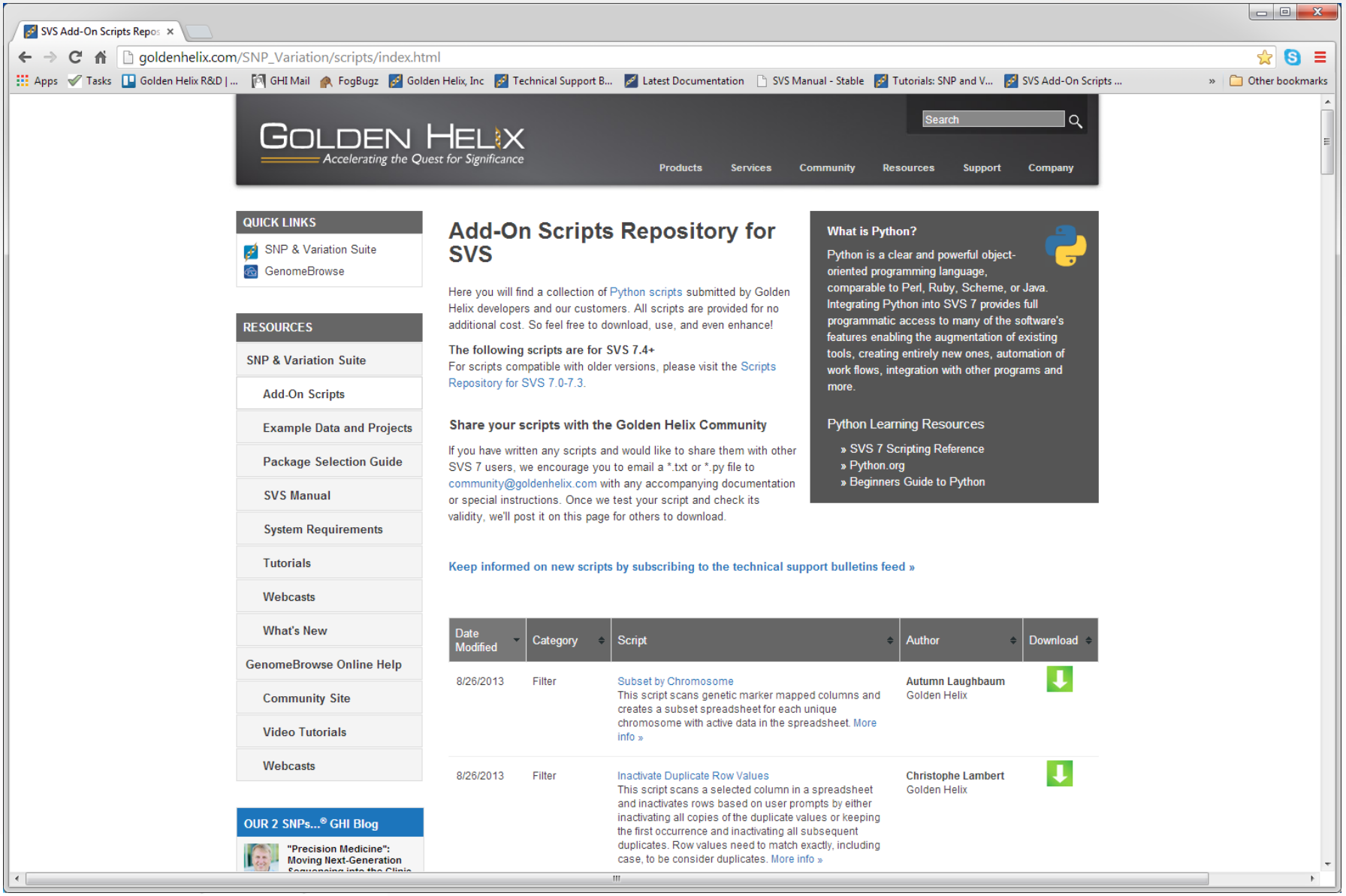
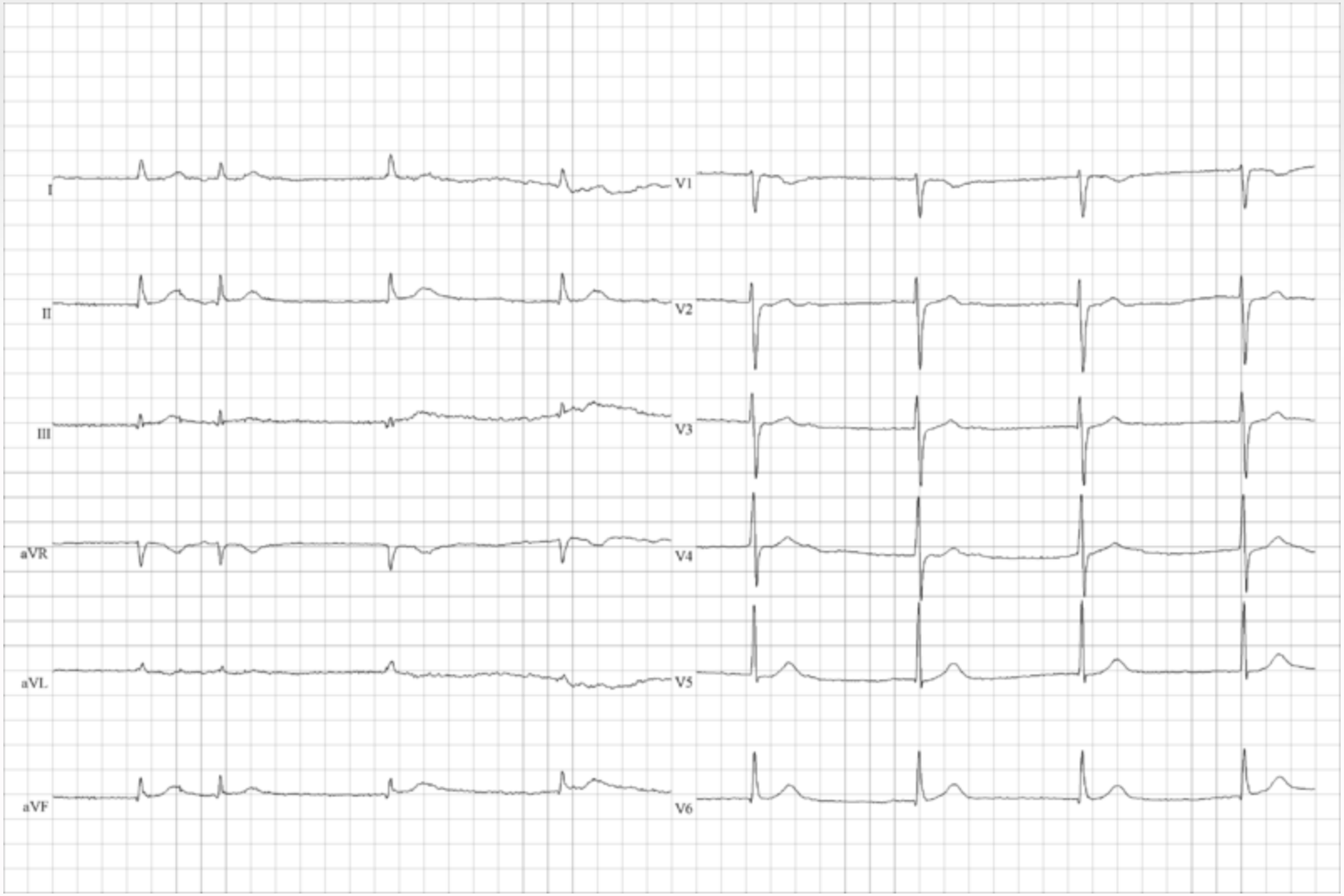
SVS Add-On Scripts Repository



SVS Demo

Exome Sequencing



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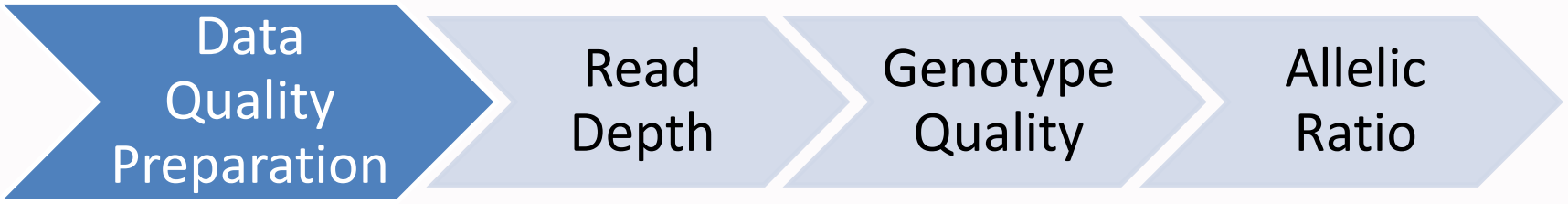
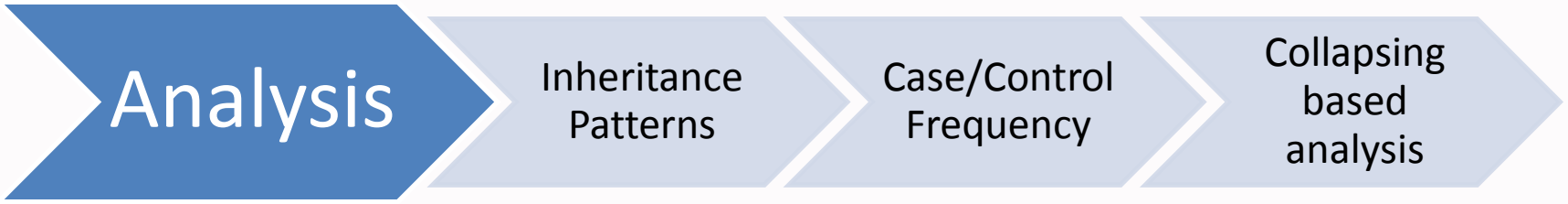

Data Quality Preparation
Read Depth
Genotype Quality
Allelic Ratio


Initial Investigation
Variant Frequencies
Functional
Damaging


Analysis
Inheritance Patterns
Case/Control Frequency
Collapsing based analysis

[Poll]

Conclusion

Questions?

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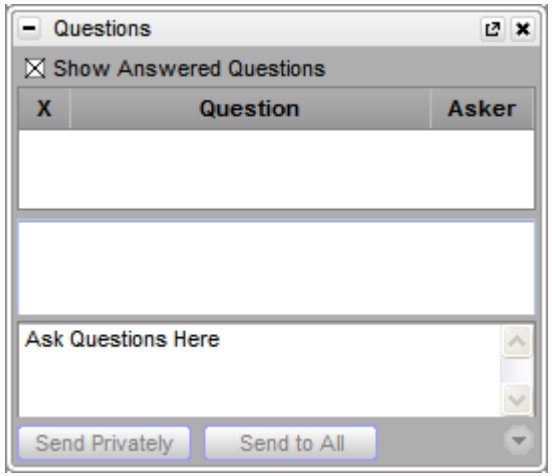

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