Example Workshop Agenda

Morning Day 1 – Introduction to SVS and Genomic Analysis Concepts

SECTION 1 – Introduction to SVS and GenomeBrowse

1. Overview of the workspace (spreadsheets, annotations, visualization, etc.)
2. Working efficiently with big data.

SECTION 2 - DNA sequencing analysis

1. Introduction to next-generation DNA sequencing analysis
2. What you need to know about "secondary" analysis
   a. Reference sequence
   b. Alignment and variant calling
   c. Variant QC
   d. File Formats
   e. Issues to be aware of
3. Tertiary or "sense making" analysis concepts
   a. Bioinformatic filtering
   b. Variant classification and annotation
   c. Functional prediction
4. Experimental design and workflow considerations

SECTION 3 – mRNA Expression Profiling

1. Sample-level normalization and transformation
2. Differential expression analysis
3. Hierarchical clustering and plotting

SECTION 4 – GWAS Overview

1. Quality Control Considerations
2. Association Testing and Regression analysis
3. Mixed linear model analysis
4. LD And Haplotype Analysis

Afternoon Day 1 – Hands-On Training with SVS

SECTION 1 – Core Features of SVS

1. Data import/export
2. Data and project management
3. Visualization
4. Principal components analysis
5. Genotype analysis
6. Regression analysis

SECTION 2 – NGS Analysis Part 1: Family-based DNA-Seq

1. Family Trios: de Novo, compound heterozygous, and autosomal recessive mutations
2. "Diagnostic Odyssey" using exome sequencing of a trio with a rare, highly penetrant disease
3. Finding the Causal Variant of a Novel X-Linked Disorder

Morning Day 2 – Hands-on Training with SVS

SECTION 3 – NGS Part 2: Advanced DNA-Seq Workflows

1. Rare variant collapsing and case-control testing
2. Somatic mutation analysis

Afternoon Day 2 – Hands-on Training with SVS

1. SNP Genome-Wide Association Study
2. mRNA Expression Profiling
3. Microarray CNV Analysis