

# SV Analysis

# 4 Main Stages

1

Biological  
Question

2

Design &  
Conduct  
Experiment

3

Prepare NGS  
Library &  
Sequence

4

Sequencing  
Analysis

Class time



# 4 Typical Stages of Variant Analysis

1

Read Quality  
Control



2

Map Reads



3

Identify Variants



4

Visualize Variants

# What is a SV

- SNV = Single Nucleotide Variant
- In reality SNV is broader than a single nucleotide.
- Better definition would be 'variant small enough that majority of reads will span the variant site(s) and map correctly on both sides of it.
- SV = Structural Variant
- Anything that is not a SNV.

# Why do we care about SVs?

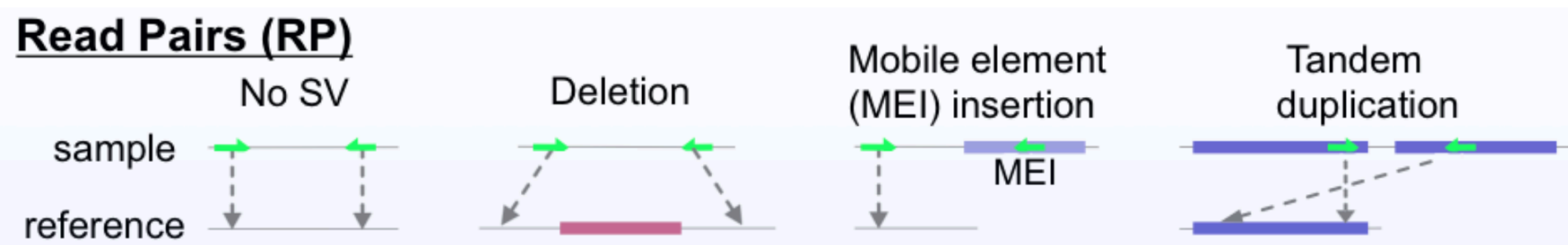
- ~25% of of total variation may be structural in nature ([Deatherage et al 2015 Frontiers in genetics](#))
- Many programs/pipelines/papers don't bother looking for them because they are classically more difficult to find.

# How do we find SVs?

- Larger insert sizes (mate pair sequencing) or read lengths (Nanopore) can significantly increase the ability to identify large rearrangements such as inversions and translocations.
- Makes use of paired end information and split read information.

# Paired end mapping

- Structural variants can cause different discordant mapping locations between pairs of reads.



# Coverage Depth

- Deletions and Amplifications are associated with decrease and increase in coverage depth respectively.
  - Coverage depth alone does not identify specific edges of deletion or amplification.
  - Particularly well hidden in mixed population data.





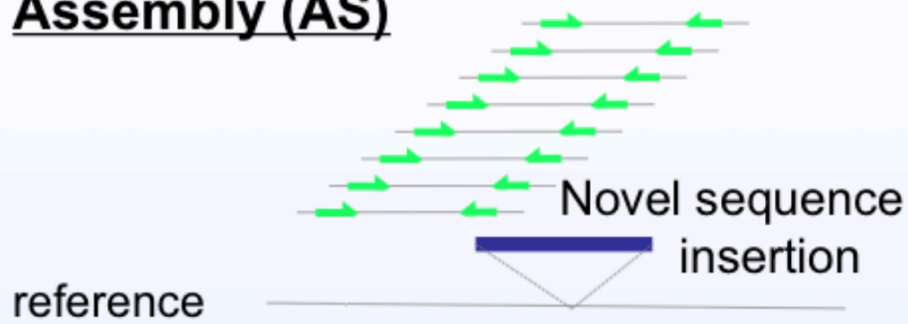
# Split read alignment

- Similar to RNA seq identifying splice variants.
- Often more powerful when leveraged with other data such as coverage data.

## Split Reads (SR)



## Assembly (AS)



# Tutorials

- Using a small data set use the Svdetect program to identify structural variants.
- This will include a quick read mapping step as it is a new data set, but try not to get bogged down on that part.
- Together with the data from the SNV tutorial tomorrow we will go through some more advanced visualization tutorials to see the variants.