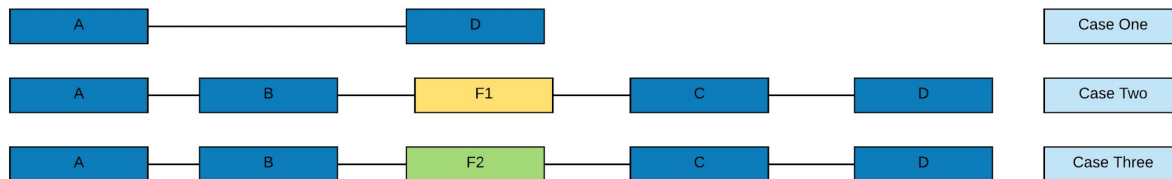
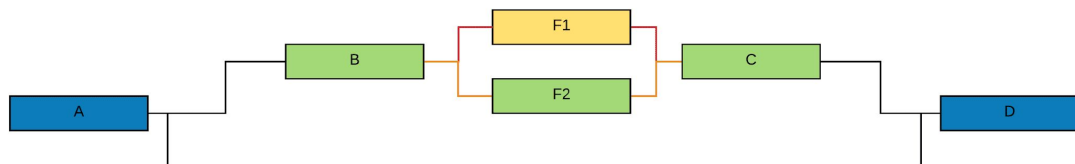
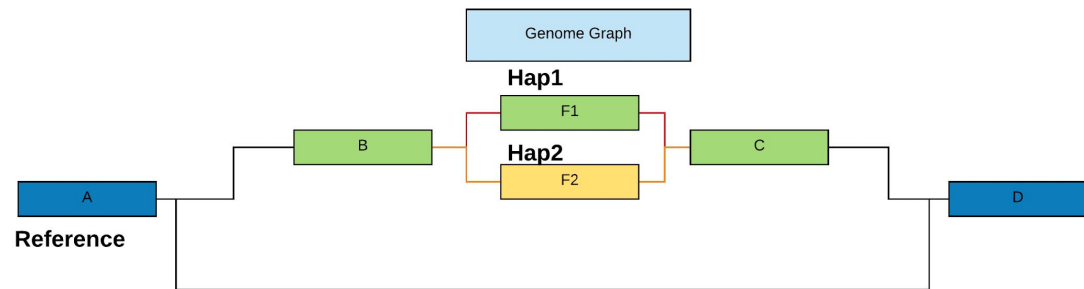


# Graph to Haplotype representation



chr21:1-100

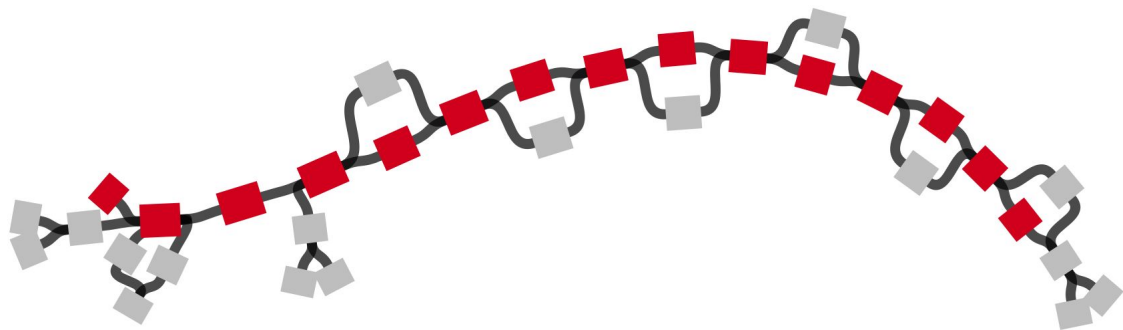
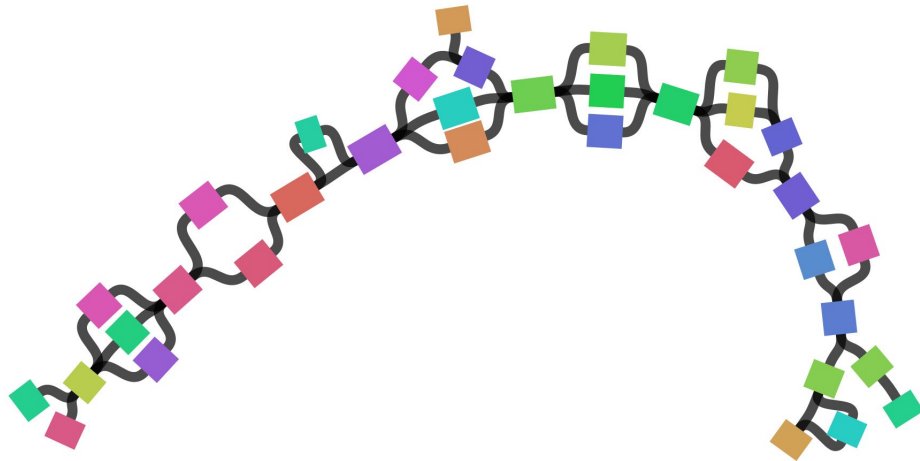
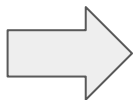
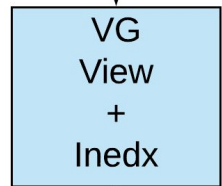
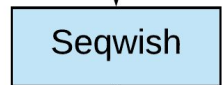
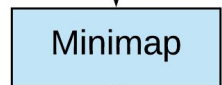
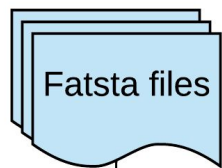
chr21:1-50>+H21\_1:1-100>+chr21:51-100

chr21:1-50>+H21\_1:1-20>+H21\_2:1-40>+H21\_1:60-100>chr21:51-100

# Rationale

chr21:1-50>+H21\_1:1-20>+H21\_2:1-40>+H21\_1:60-100>chr21:51-100

- Ability to add new haplotype without altering existing reference coordinate
- Reused part of shared sequence among haplotypes (partial overlap, inversion)
- Support patch of reference genome
- Ability to swap out and create custom reference based on preferable haplotype.



# How to add new haplotype?

- First come first serve, but major haplotype if possible
- No breaking existing haplotype
- Build new Hap based on the closet Hap from MSA
  - Chain file indication junction of haplotype
  - Reuse prefix/suffix of existing haplotype

# When to add new haplotype?

- Sufficient shared allele frequency ( $>1\%$ ) and number of individual ( $>10k$  people)
- Significant alteration (X% mismatch/indel/ N bp missing)
  - Caveat: different mapping would have different sensitivity to handle diverged sequence