

PCAWG-6
SV Merging
Nov 2015 releases

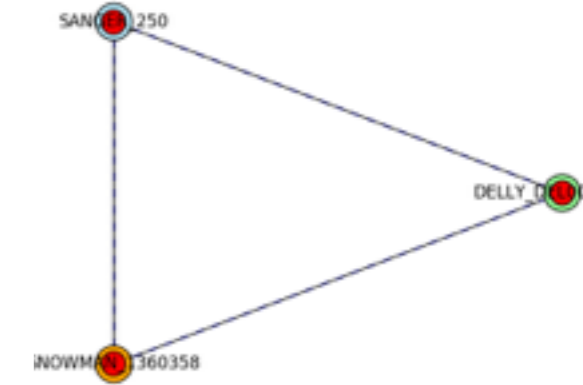
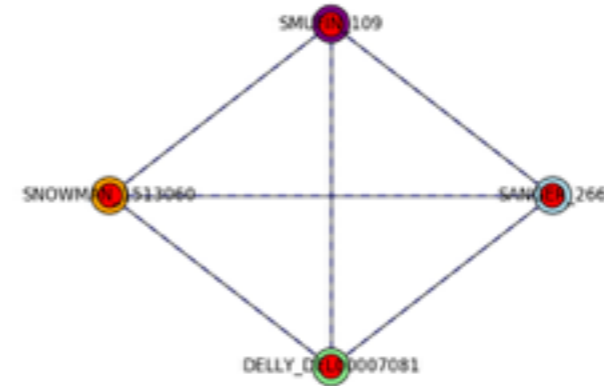
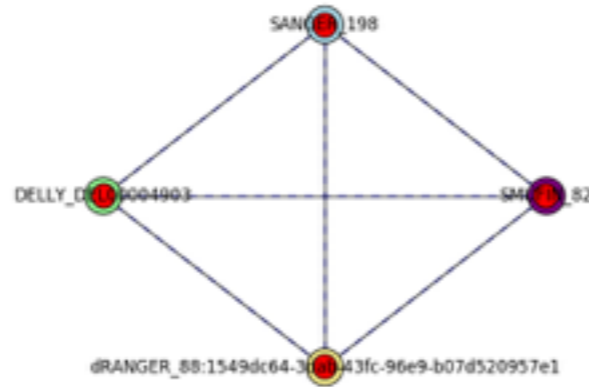
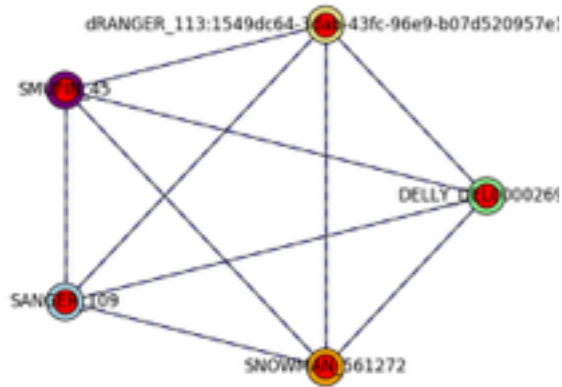
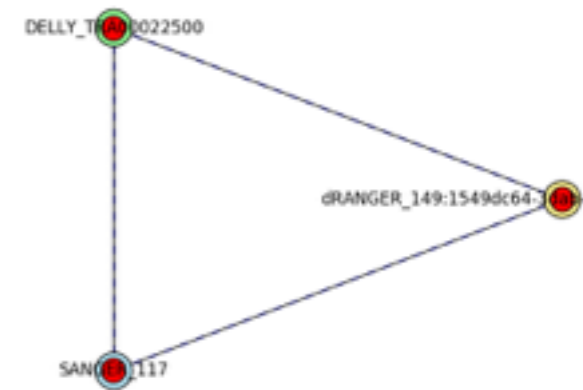
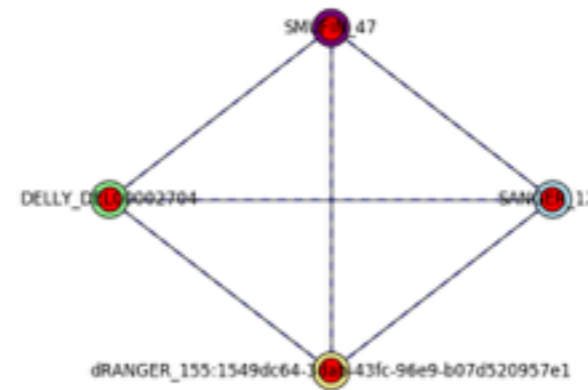
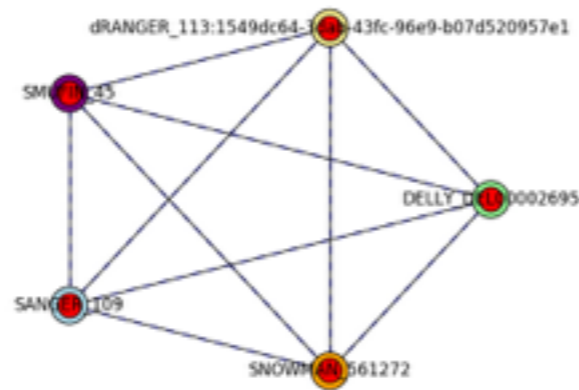
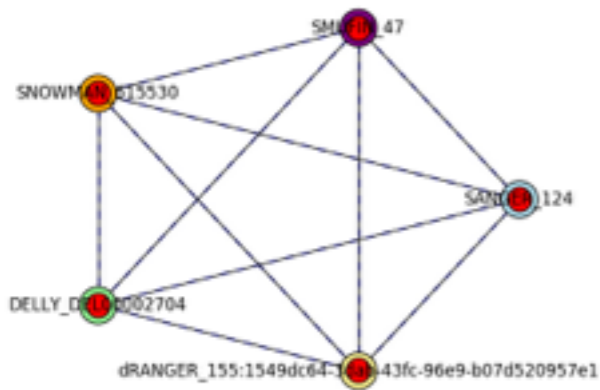
Joachim Weischenfeldt
EMBL/BRIC

Compare and merge SV calling pipelines to identify sensible metrics for merging

- SV calls from Autumn/winter releases:
 - BRASS (Sanger, Oct 2015)
 - DELLY (EMBL, Jan 2016)
 - dRANGER (BROAD, Oct 2015)
 - SMUFIN (BSC, Dec 2015)
 - SNOWMAN/dRANGER merge (BROAD, Dec 2015)
 - Currently, 132 samples in common (>2000 samples soon available for all)

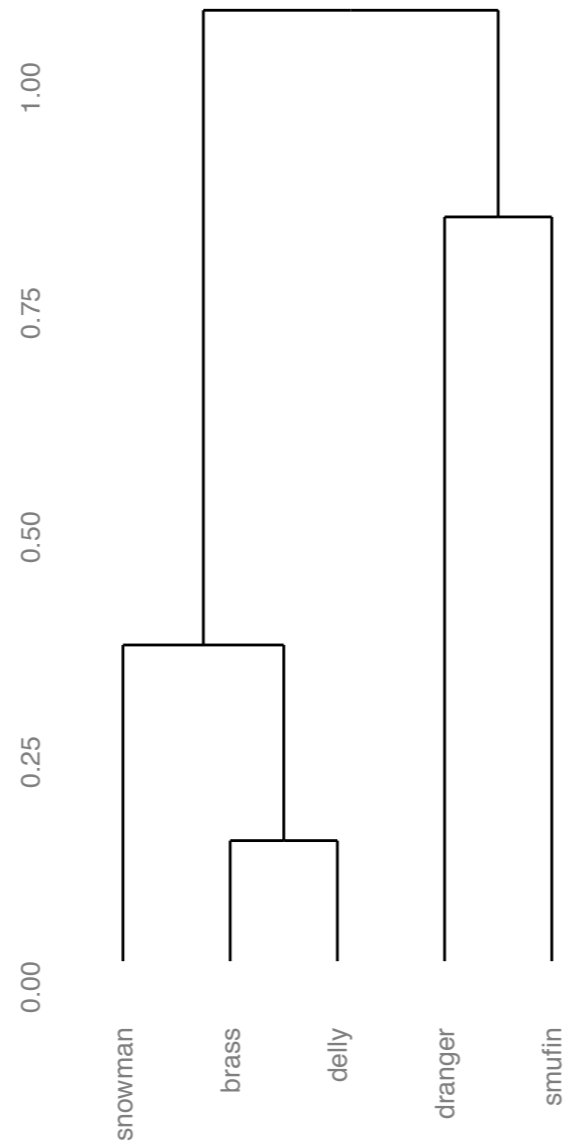
Finding 'cliques' of SVs

due to missing read group info and read quality, this criteria was not included

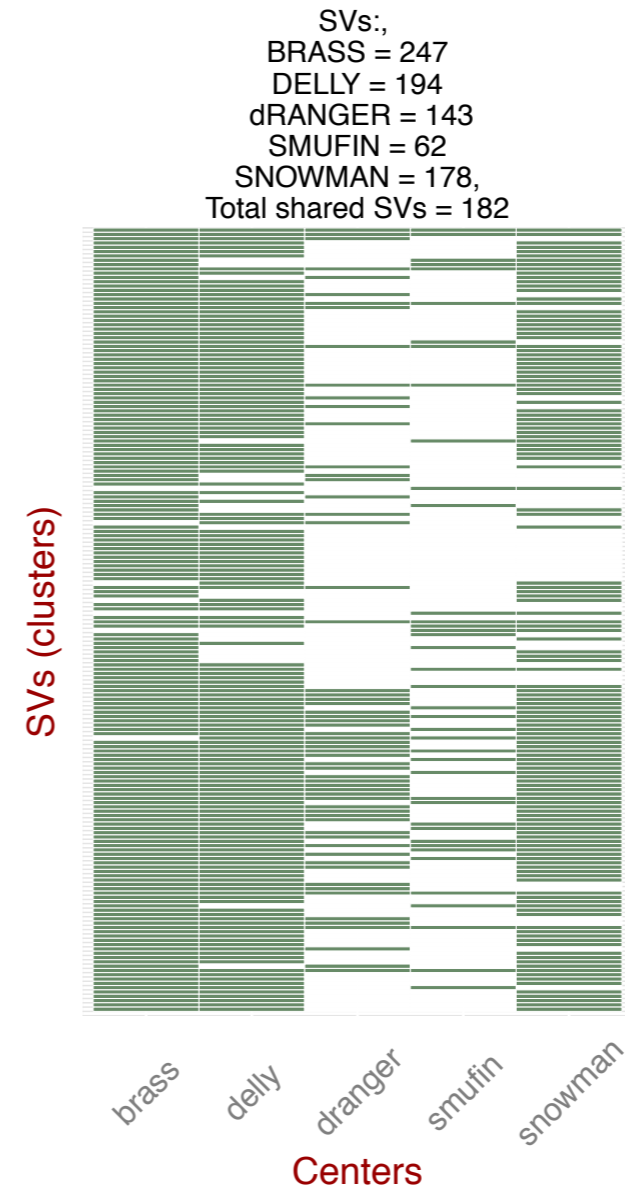


BLCA

PCAWG SV caller clustering
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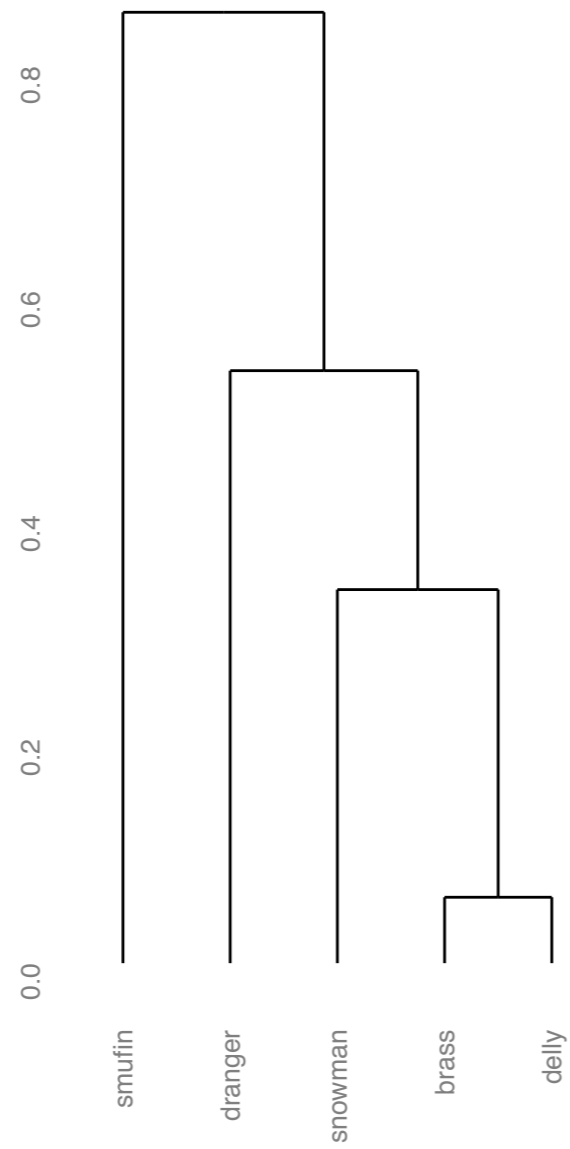


PCAWG SV concordance

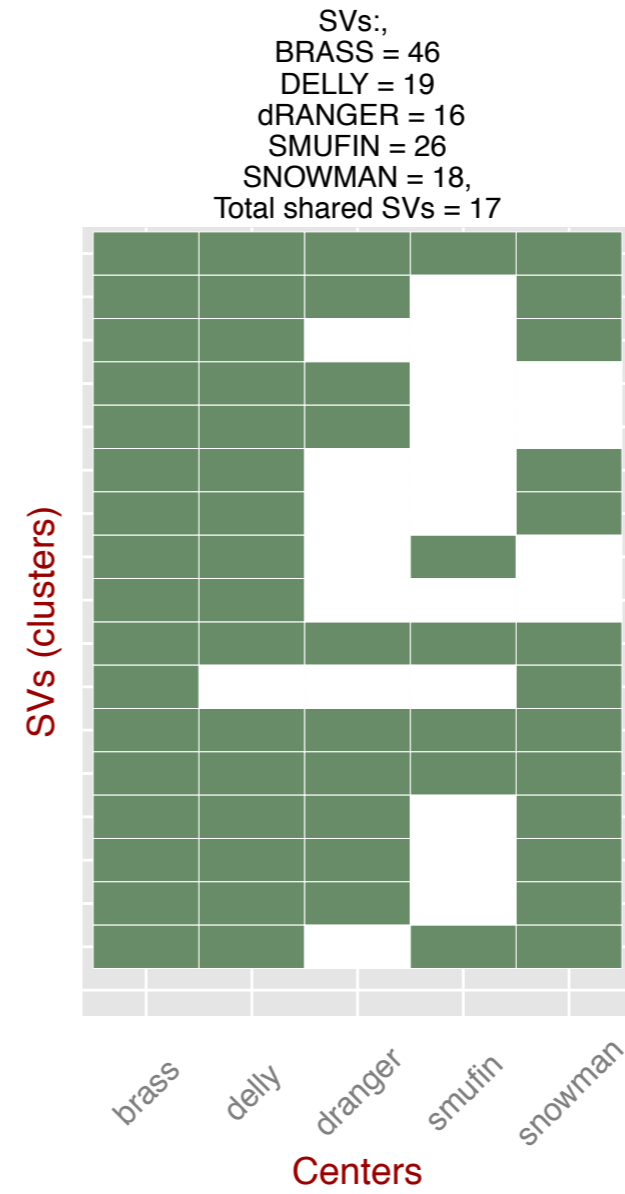


BLCA

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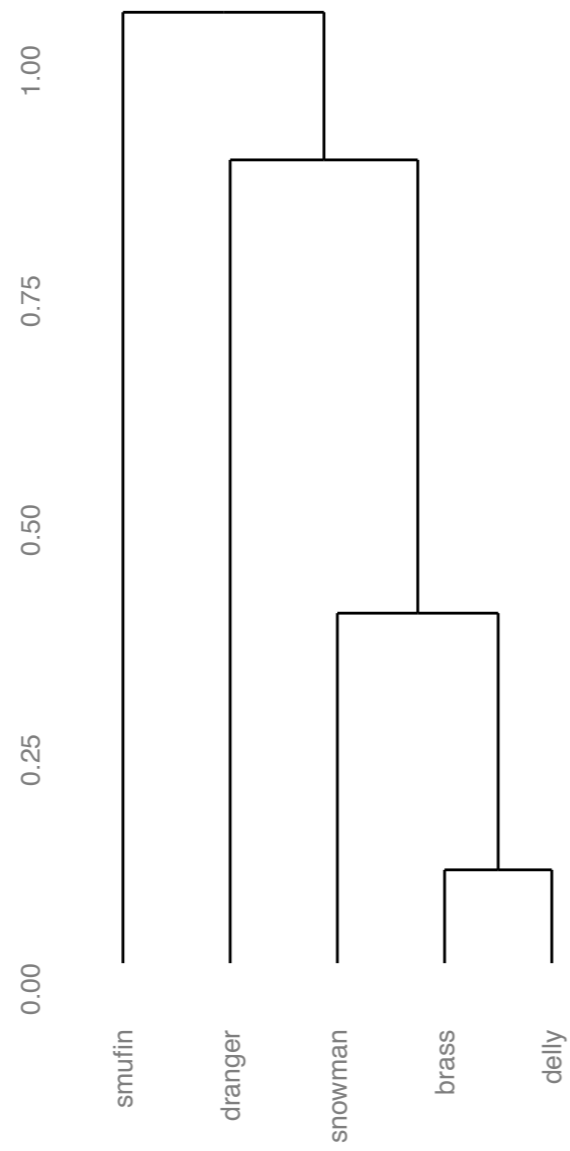


PCAWG SV concordance

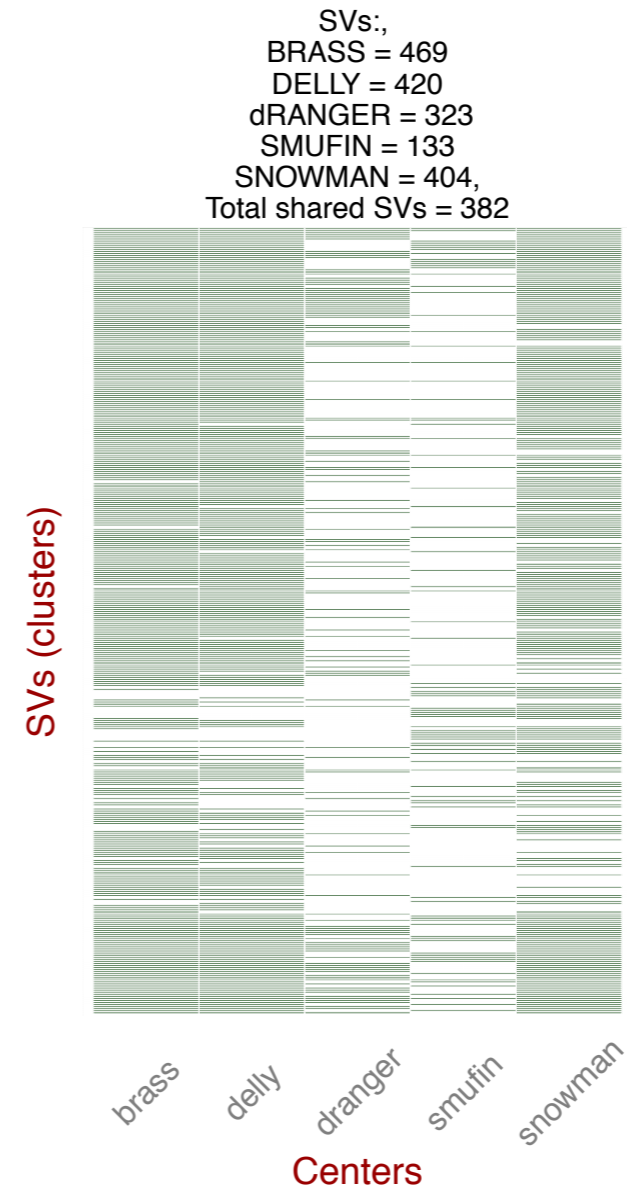


BLCA

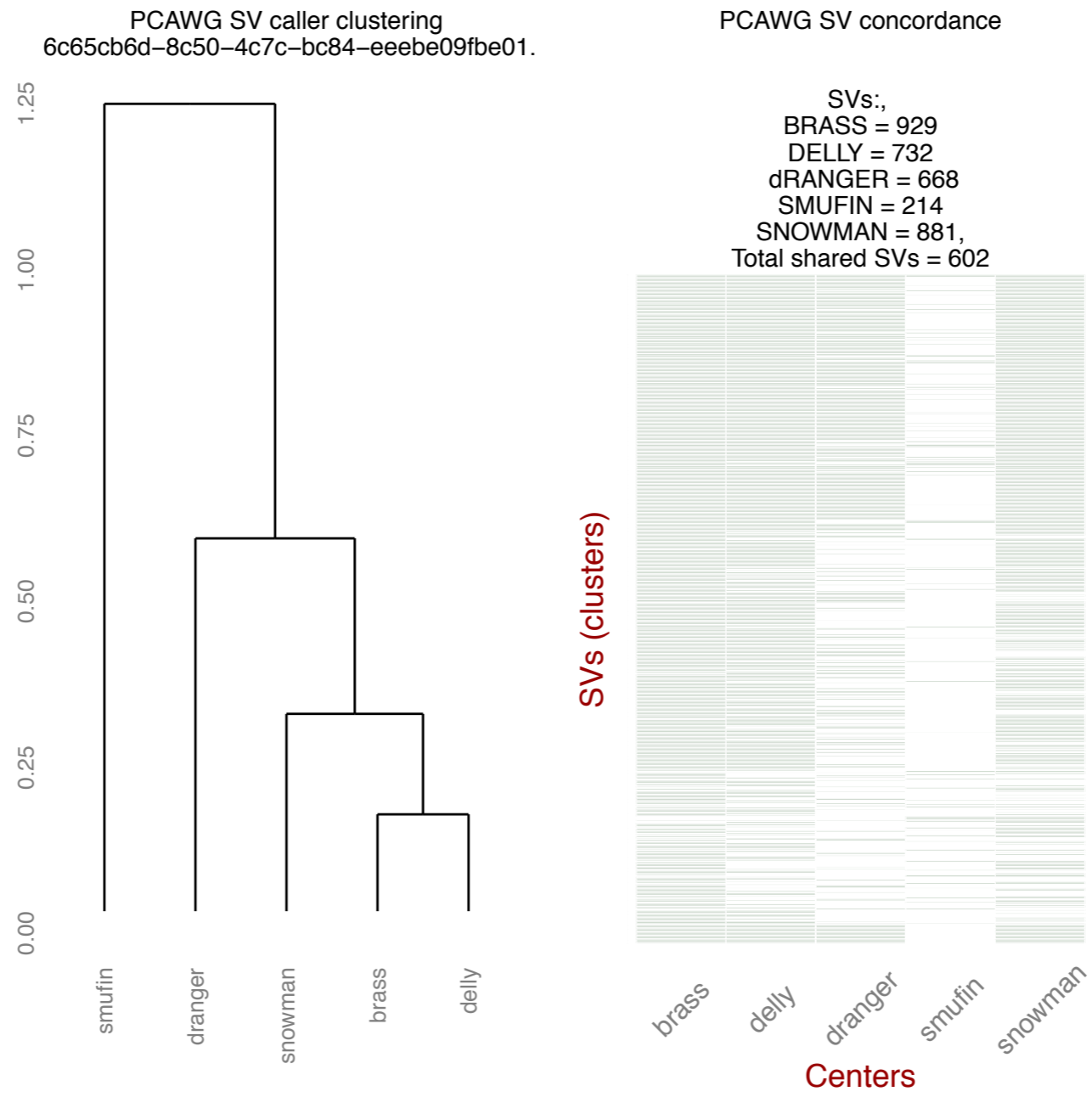
PCAWG SV caller clustering
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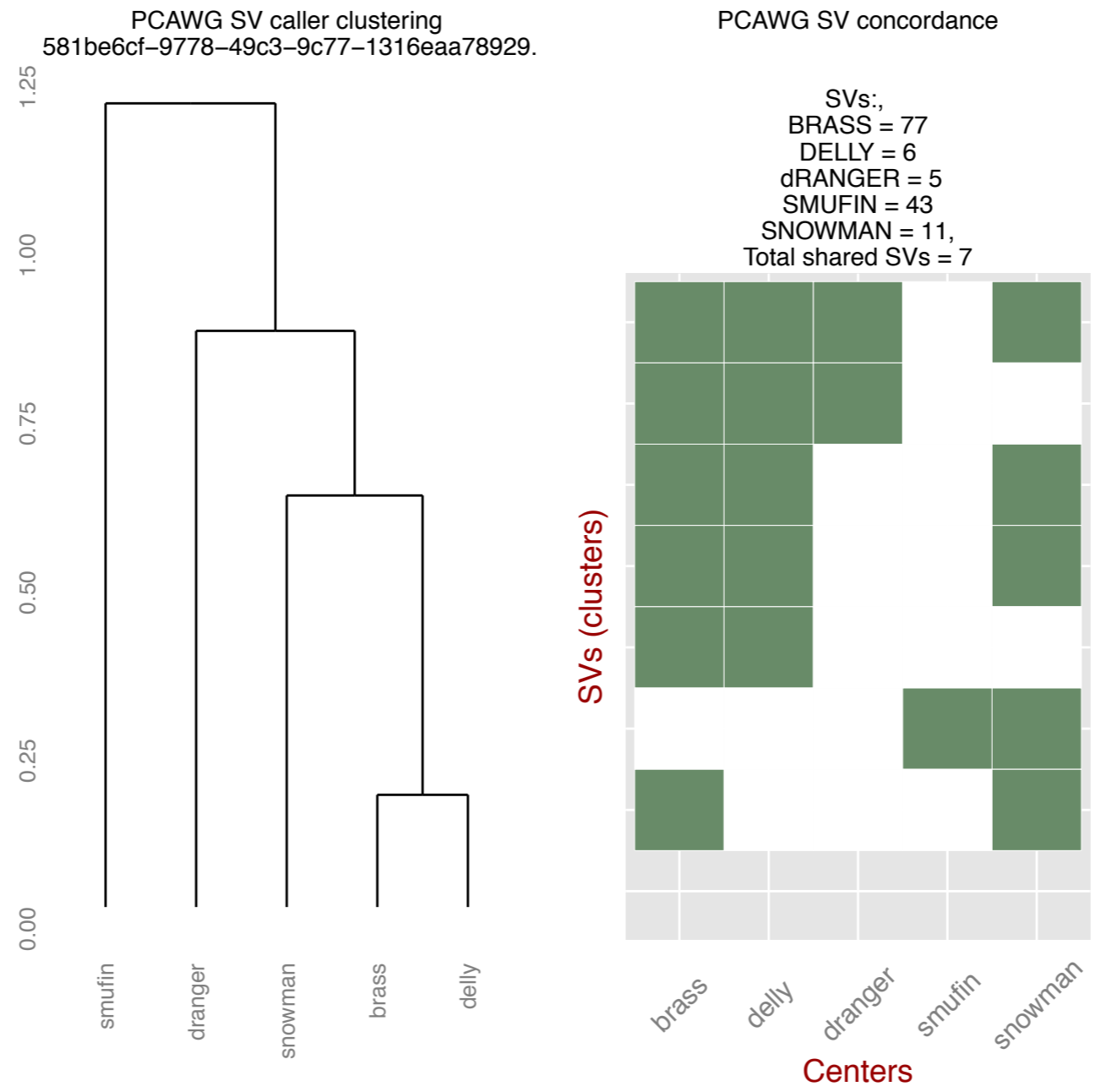
PCAWG SV concordance



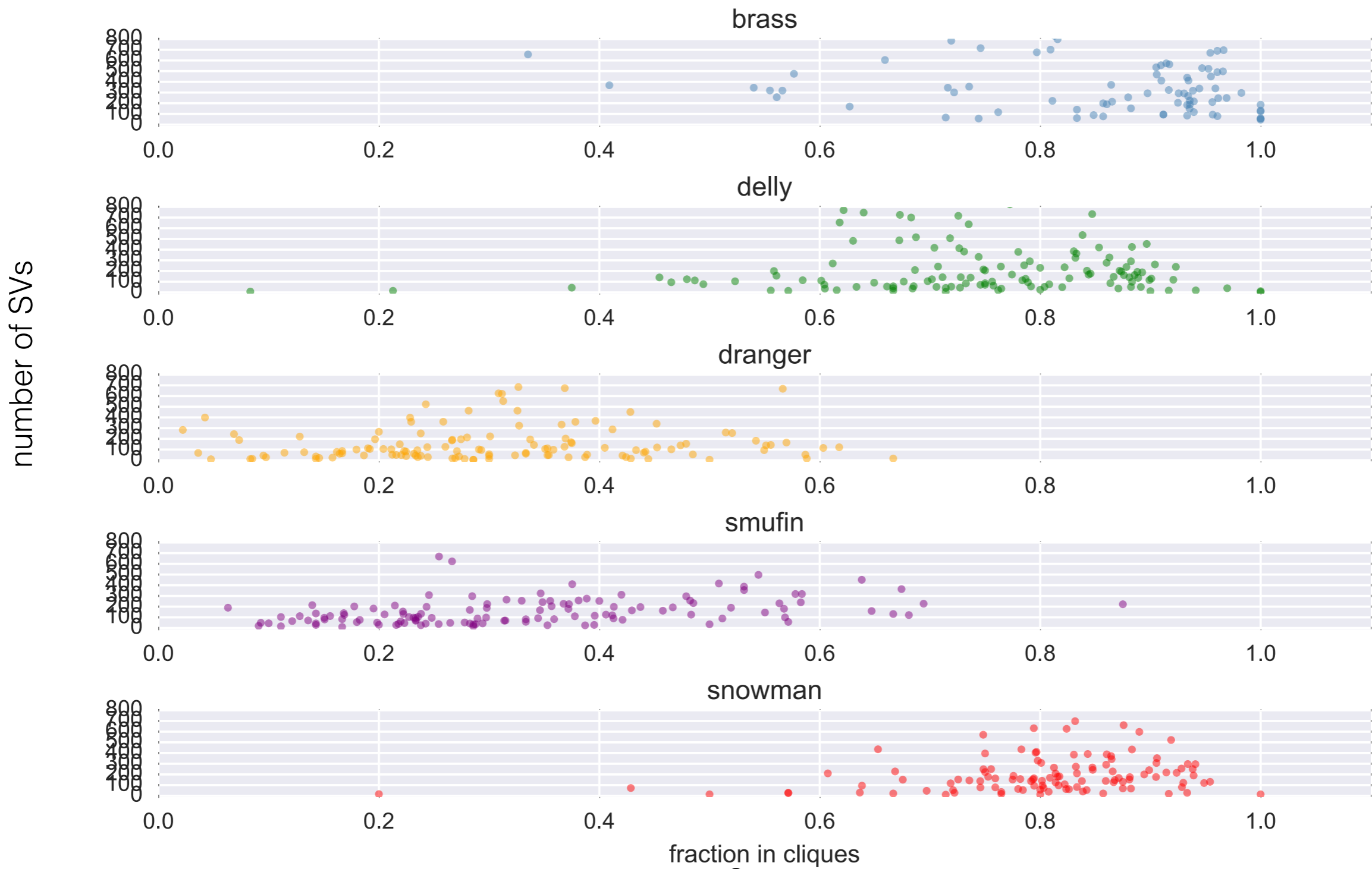
BRCA



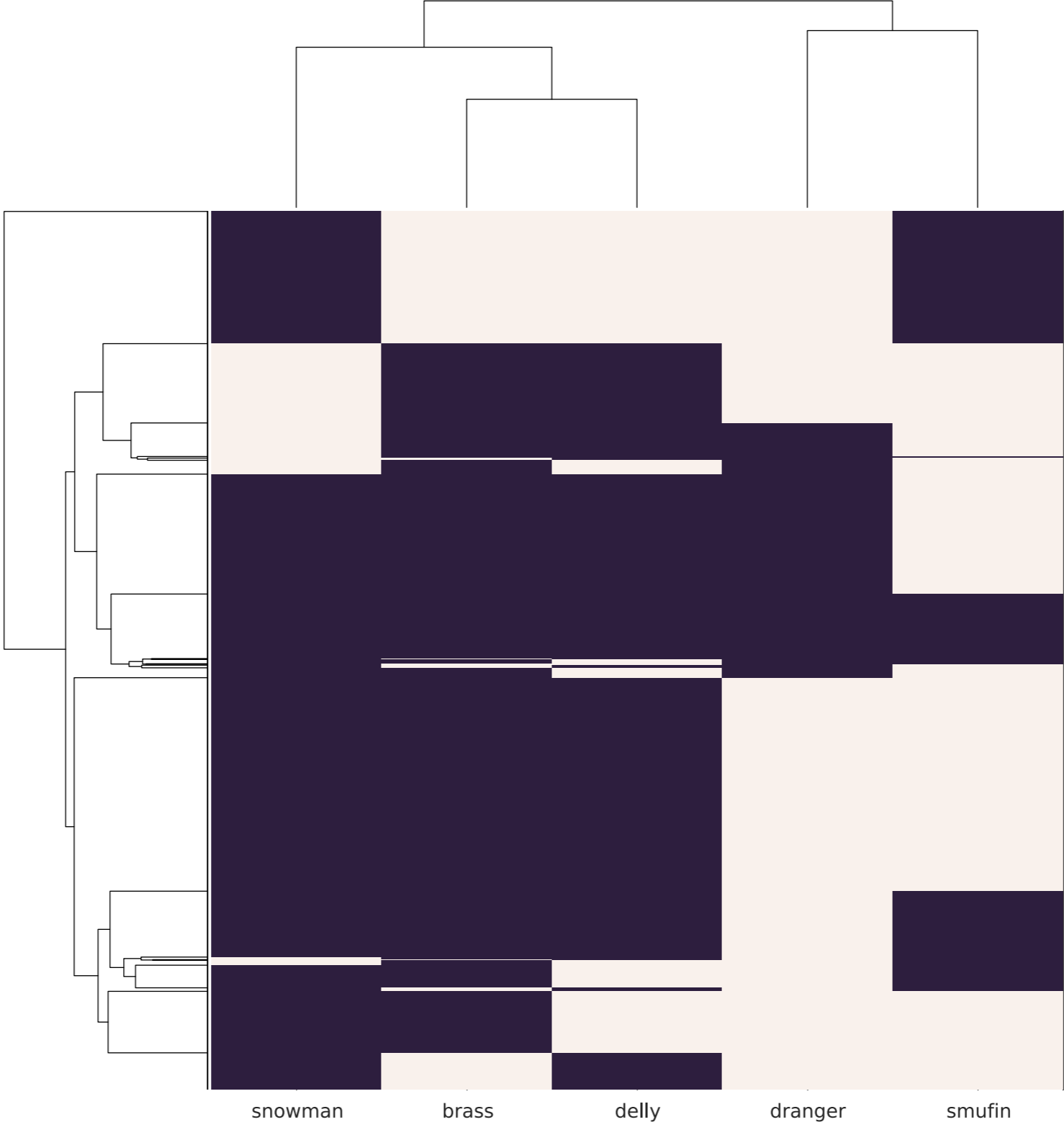
PRAD



partitioning SV callers into 'fraction in cliques'



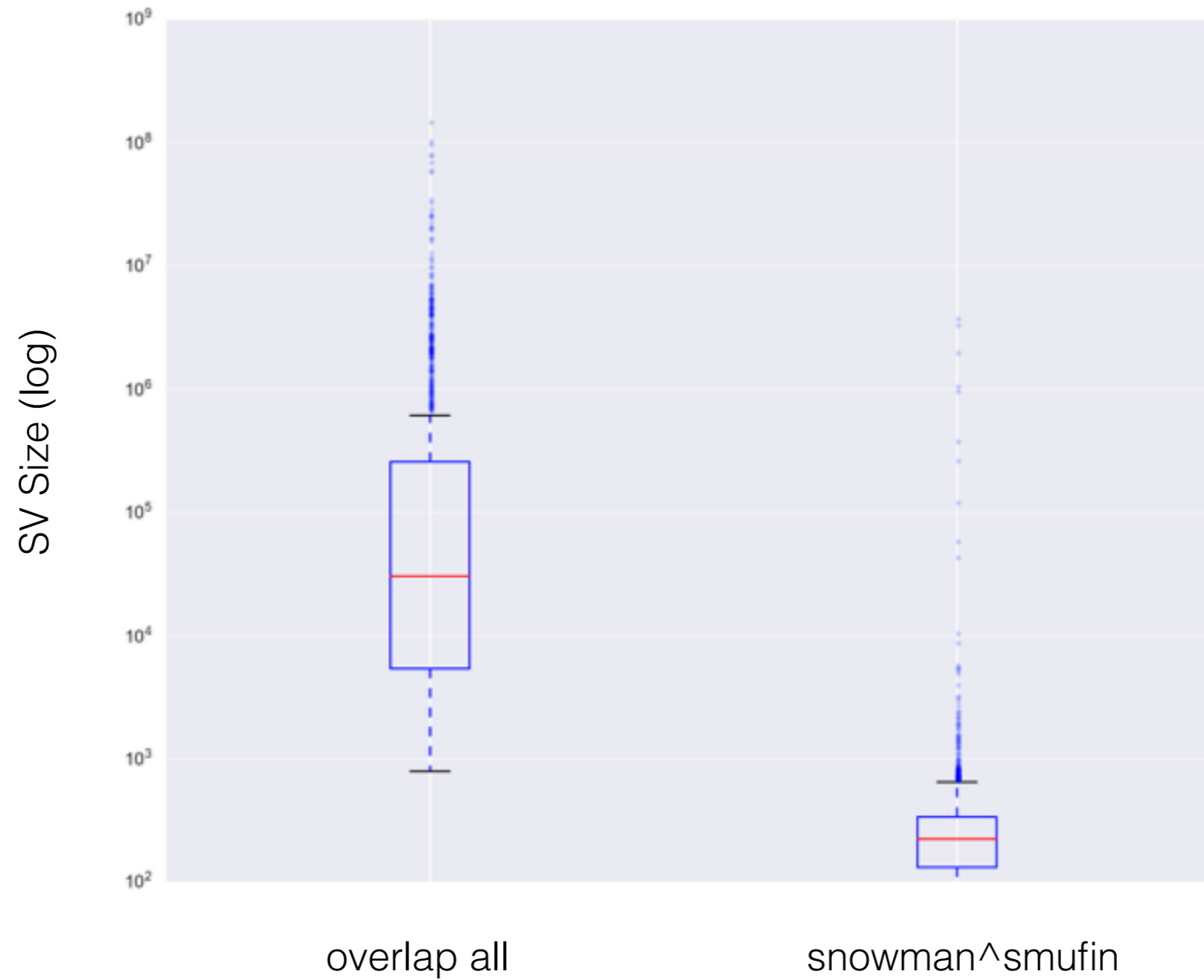
Clustering of SV callers based on 4,000 random SV cliques (out of 22,000 cliques from 132 tumor samples)



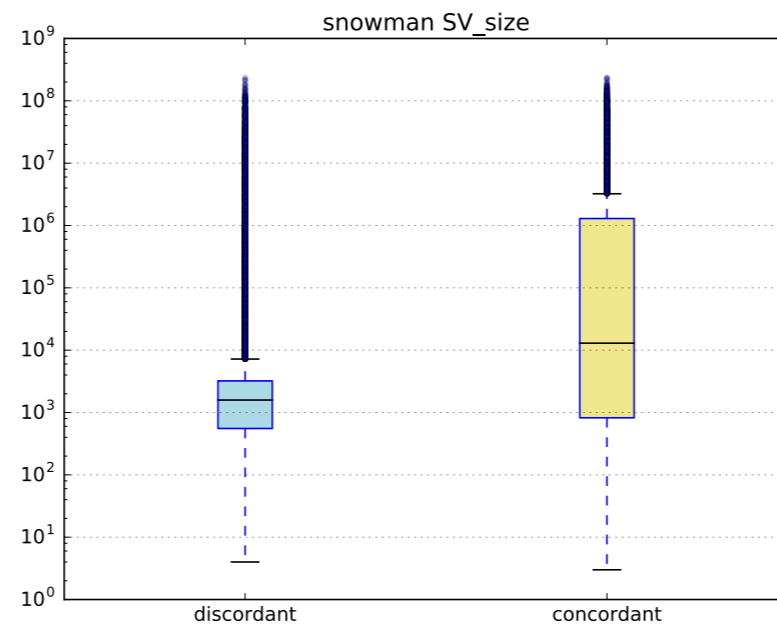
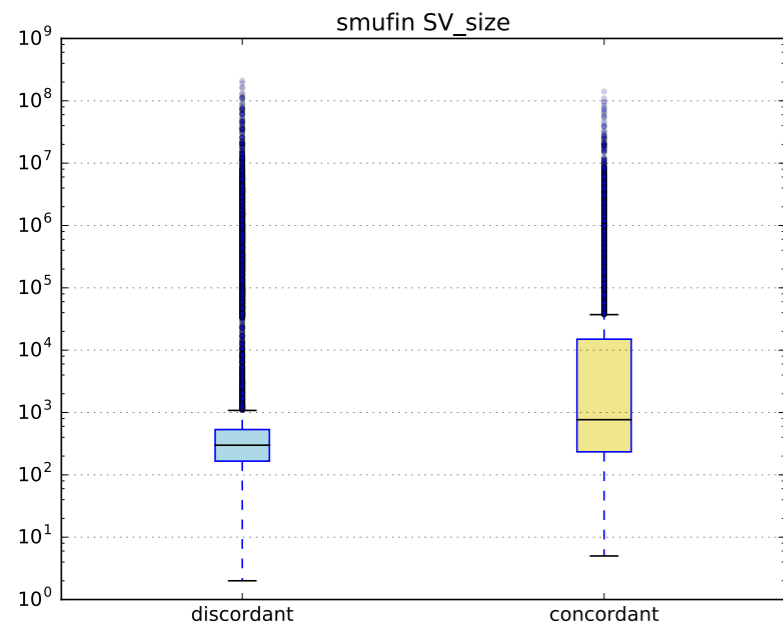
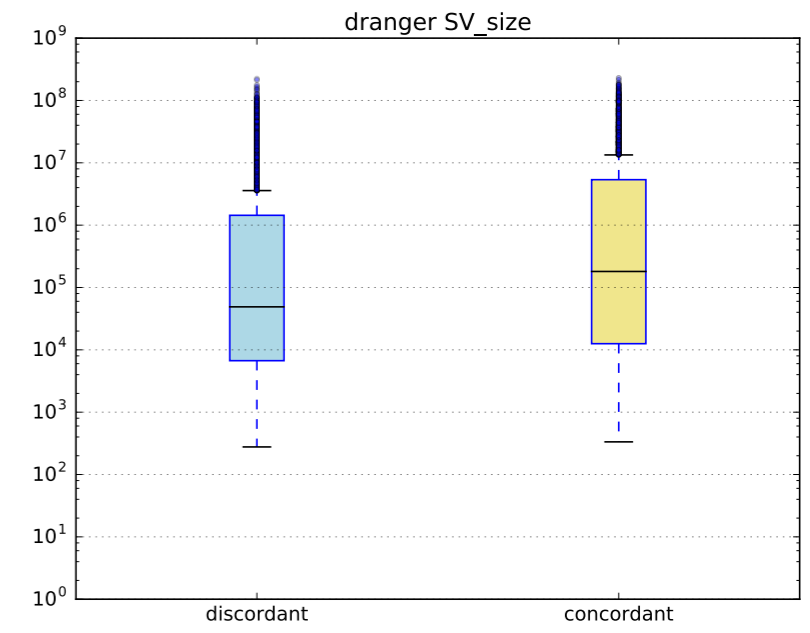
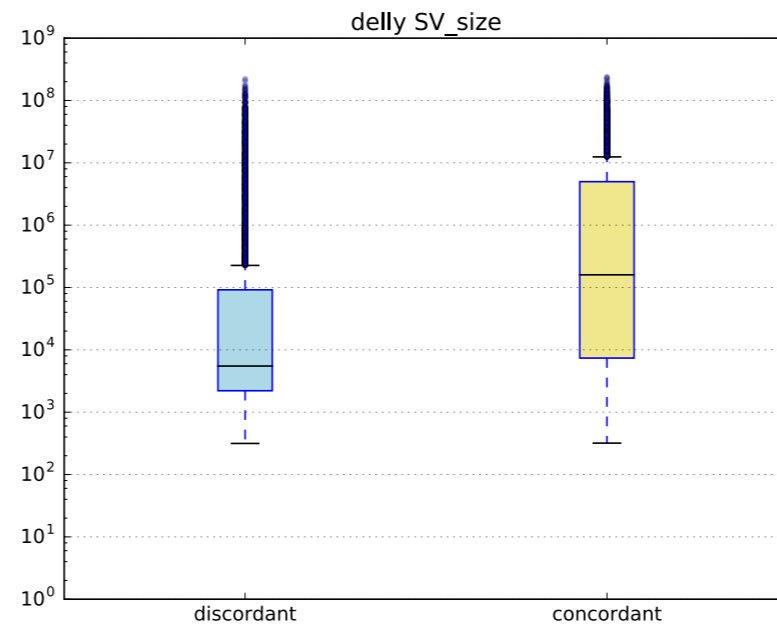
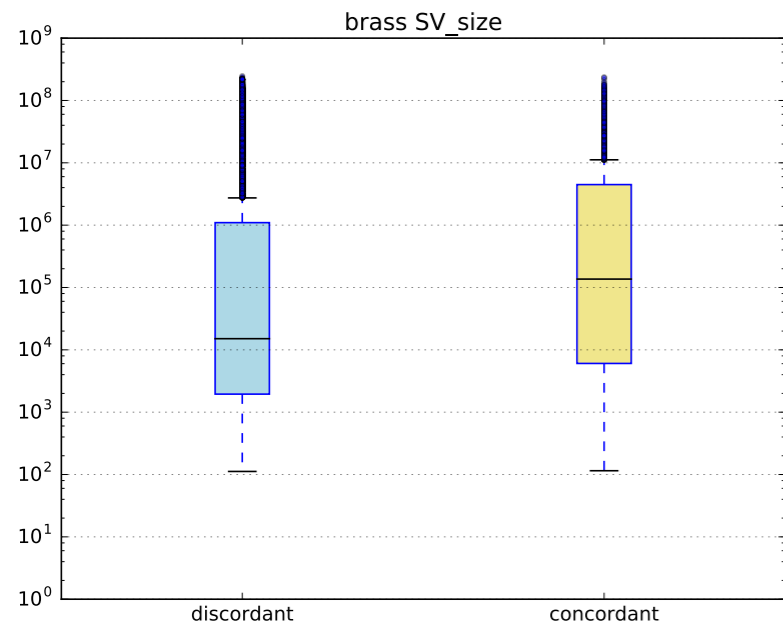
pairwise correlation of SV callers



smufin and snowman share small size SVs



SV Size



- ToDo:
 - intersect with copy number changes (jabba)
 - assess type and location of private SVs - read depth plots/IGV for manual inspection (ongoing)