Why are we aggregating

- What is our point here
  - H1: SVs cause SNPs (BMR shift)?
  - H2: SNPs cause SVs
  - H3: there is another factor that causes both;

- Order matters: What happened first what happened later?

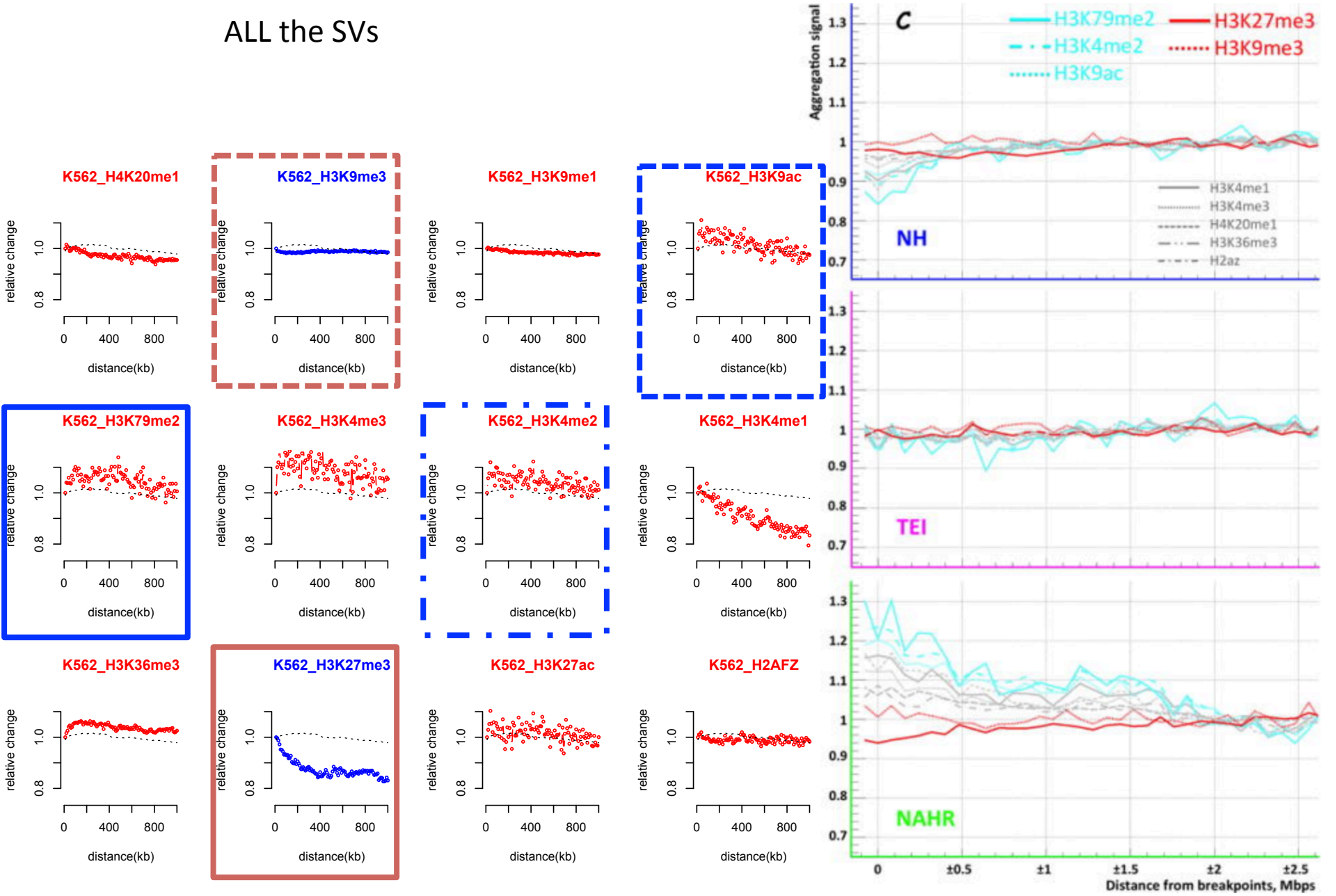
- Germline common SVs + germline rare SNVs?
  - Adjusted for ethnicities/subpopulation
- Germline common SVs + any cancer SNVs?
- Somatic SVs + SNVs (probably mostly co-occurrence)

Which call set for Gm12878

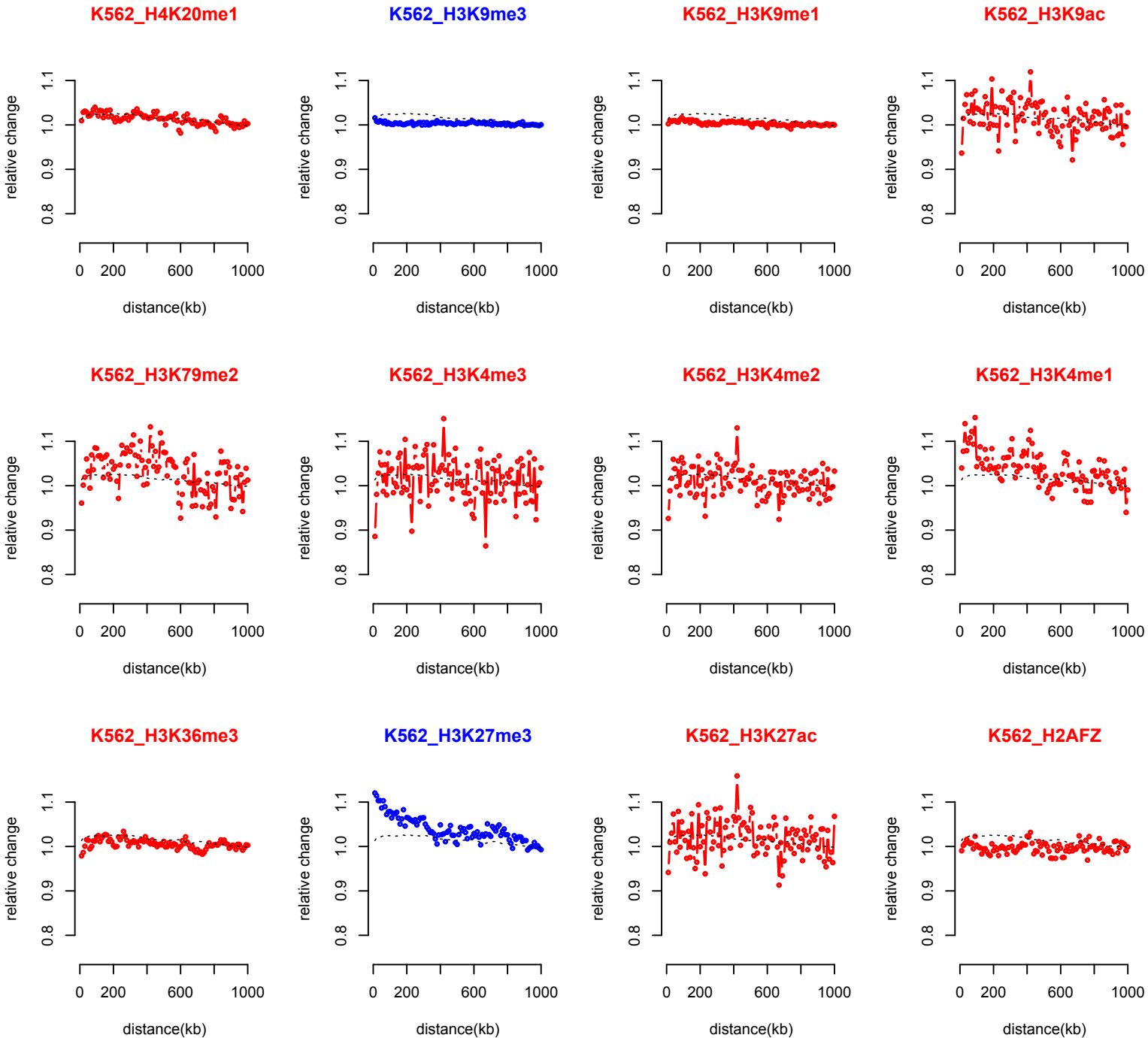
- Genome in a bottle (NIST)?
- 2014 Lumpy paper (high confidence)
- 2018 Nanopore…probably not great for SVs

- 1000 Genome?

ALL the SVs


K562_H4K20me1
K562_H3K9me3
K562_H3K9me1
K562_H3K9ac
K562_H3K79me2
K562_H3K4me3
K562_H3K4me2
K562_H3K4me1
K562_H3K36me3
K562_H3K27me3
K562_H3K27ac
K562_H2AFZ
relative change
distance(kb)
0
400
800
0.8
1.0
C
H3K79me2
H3K27me3
H3K4me2
H3K9me3
H3K9ac
H3K4me1
H3K4me3
H4K20me1
H3K36me3
H2az
Aggregation signal
NH
TEI
NAHR
0.7
0.8
0.9
1
1.1
1.2
1.3
±0.5
±1
±1.5
±2
±2.5
Distance from breakpoints, Mbps

Deletions only



Insertion only