

1) Zhou .. Urban paper SVs

5490
 531 <DUP:TANDEM>
 2602 <INS>
 436 <INV>
 23 <UNK>

2) From XK: enhancers overlapping SVs & genes nearest to the enhancers

192 DEL__enhancer_loss
 23 INS__enhancer_gain

3) Using DL's linkage data

189 DEL__closest_enhancer_loss
 3 DEL__linkage_enhancer_loss
 23 INS__closest_enhancer_gain

4) Target genes w SVs in enhancers in DE analysis (> 1 total read count across all samples)

41 DEL__closest_enhancer_loss
 3 DEL__linkage_enhancer_loss
 6 INS__closest_enhancer_gain

5) Ploidy and CN correction

Zhou .. Urban CNVs overlapped w genes by STL

| all genes | | genes w SV in enhancer | |
|-----------|-----|------------------------|-----|
| 95 | CN0 | | CN0 |
| 464 | CN1 | | CN1 |
| 13782 | CN2 | 18 | CN2 |
| 32060 | CN3 | 21 | CN3 |
| 9833 | CN4 | 11 | CN4 |
| 1022 | CN5 | | CN5 |

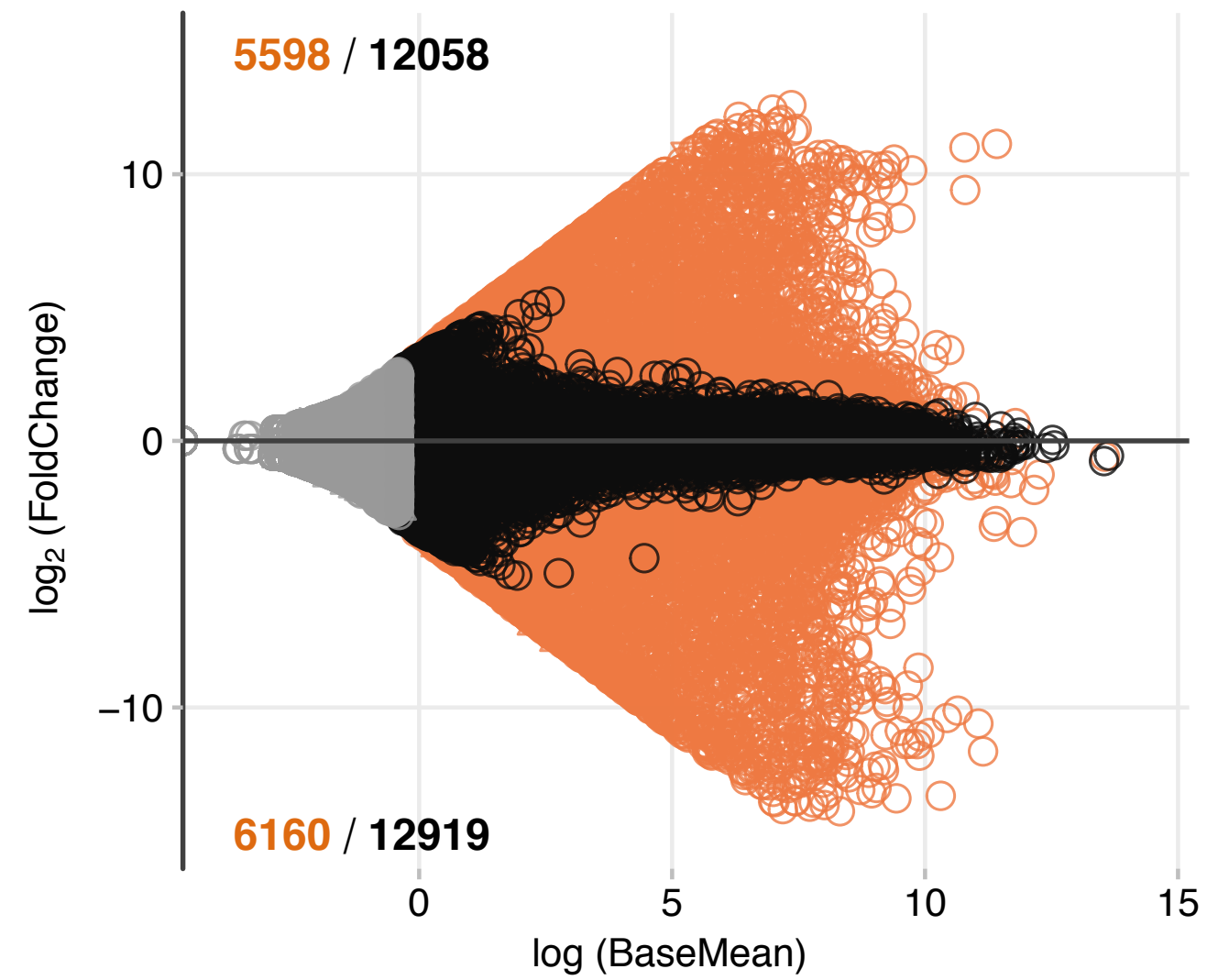
adjusted p-value:

- > 0.05
- NA
- ≤ 0.05

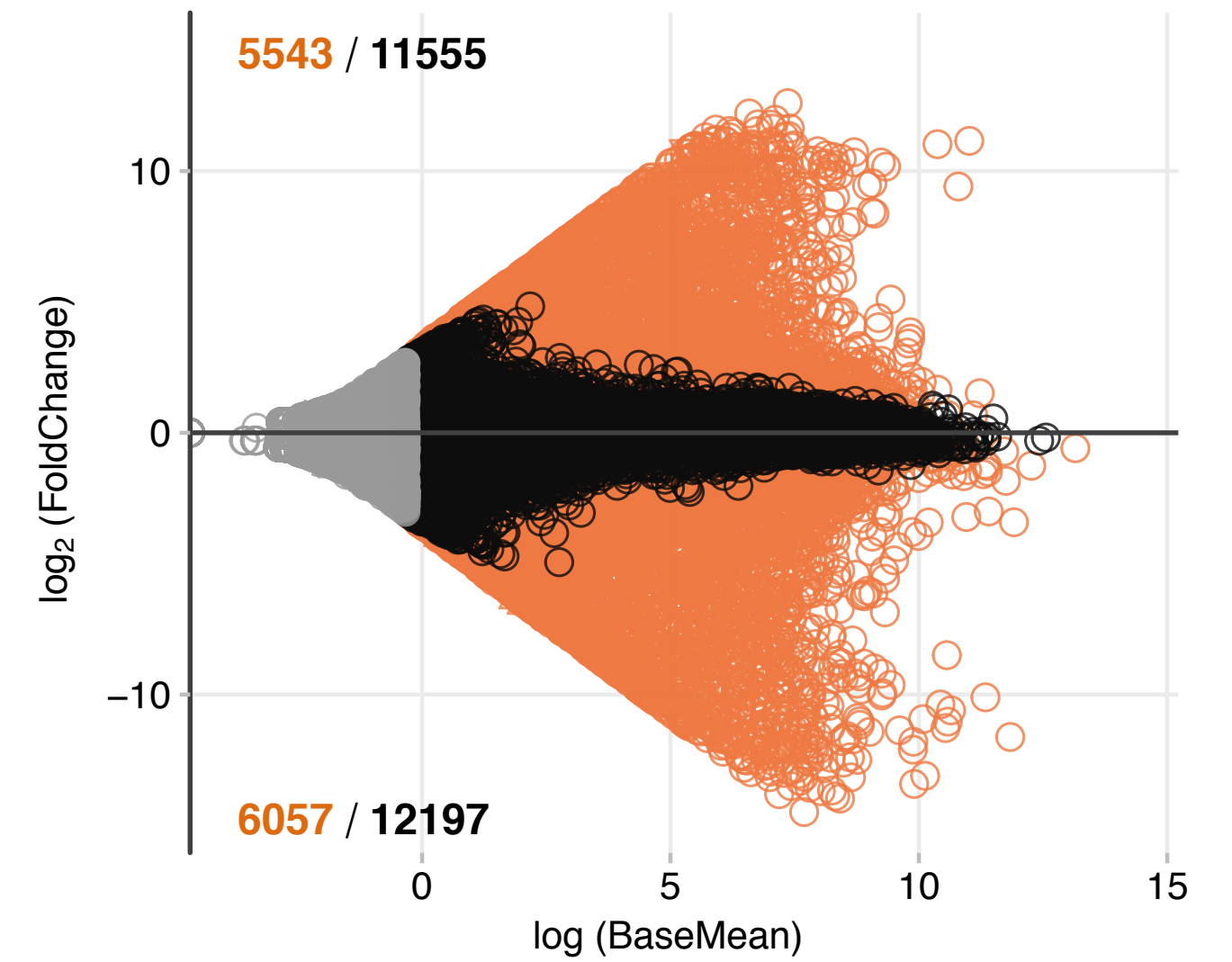
type of K562 SV, linkage, and effect:

- △ DEL__closest_enhancer_loss
- ▲ DEL__linkage_enhancer_loss
- ▽ INS__closest_enhancer_gain
- none

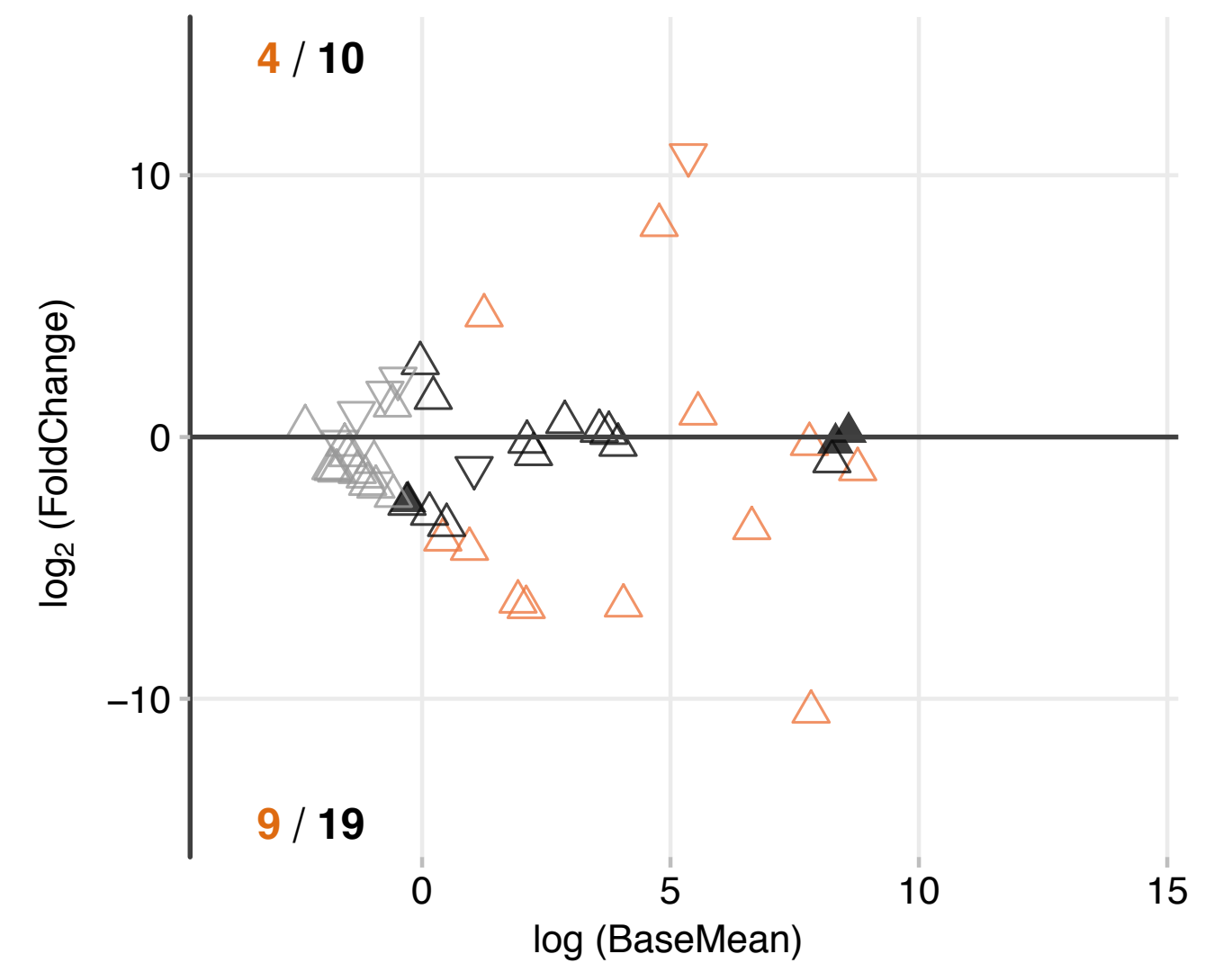
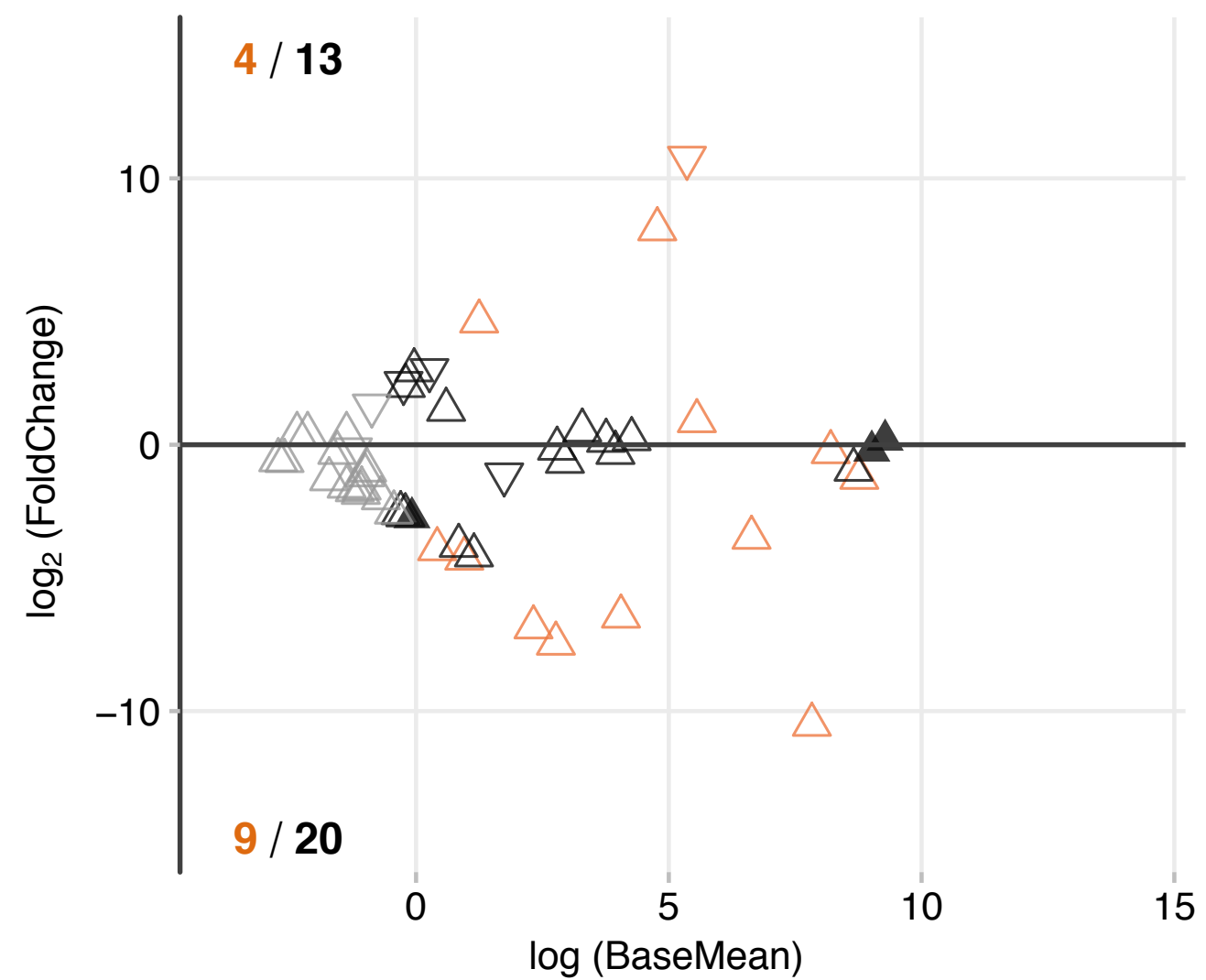
K562 vs GM12878, all genes MA plot



.. based on CN adjusted counts



only genes w K562 SVs in enhancer



1) Feng Yue lab SVs

1056 deletion
 735 insertion
 280 interchr_translocation
 44 intrachr_translocation
 66 inversion

2) From XK: enhancers overlapping SVs & genes nearest to the enhancers

126 DEL__enhancer_loss
 6 INS__enhancer_gain

3) Using DL's linkage data

121 DEL__closest_enhancer_loss
 5 DEL__linkage_enhancer_loss
 6 INS__closest_enhancer_gain

4) Target genes w SVs in enhancers in DE analysis (> 1 total read count across all samples)

30 DEL__closest_enhancer_loss
 2 DEL__linkage_enhancer_loss
 2 INS__closest_enhancer_gain

5) Ploidy and CN correction

Zhou .. Urban CNVs overlapped w genes by STL

| all genes | | genes w SV in enhancer | |
|-----------|-----|------------------------|-----|
| 95 | CN0 | | CN0 |
| 464 | CN1 | | CN1 |
| 13782 | CN2 | 10 | CN2 |
| 32060 | CN3 | 20 | CN3 |
| 9833 | CN4 | 4 | CN4 |
| 1022 | CN5 | | CN5 |

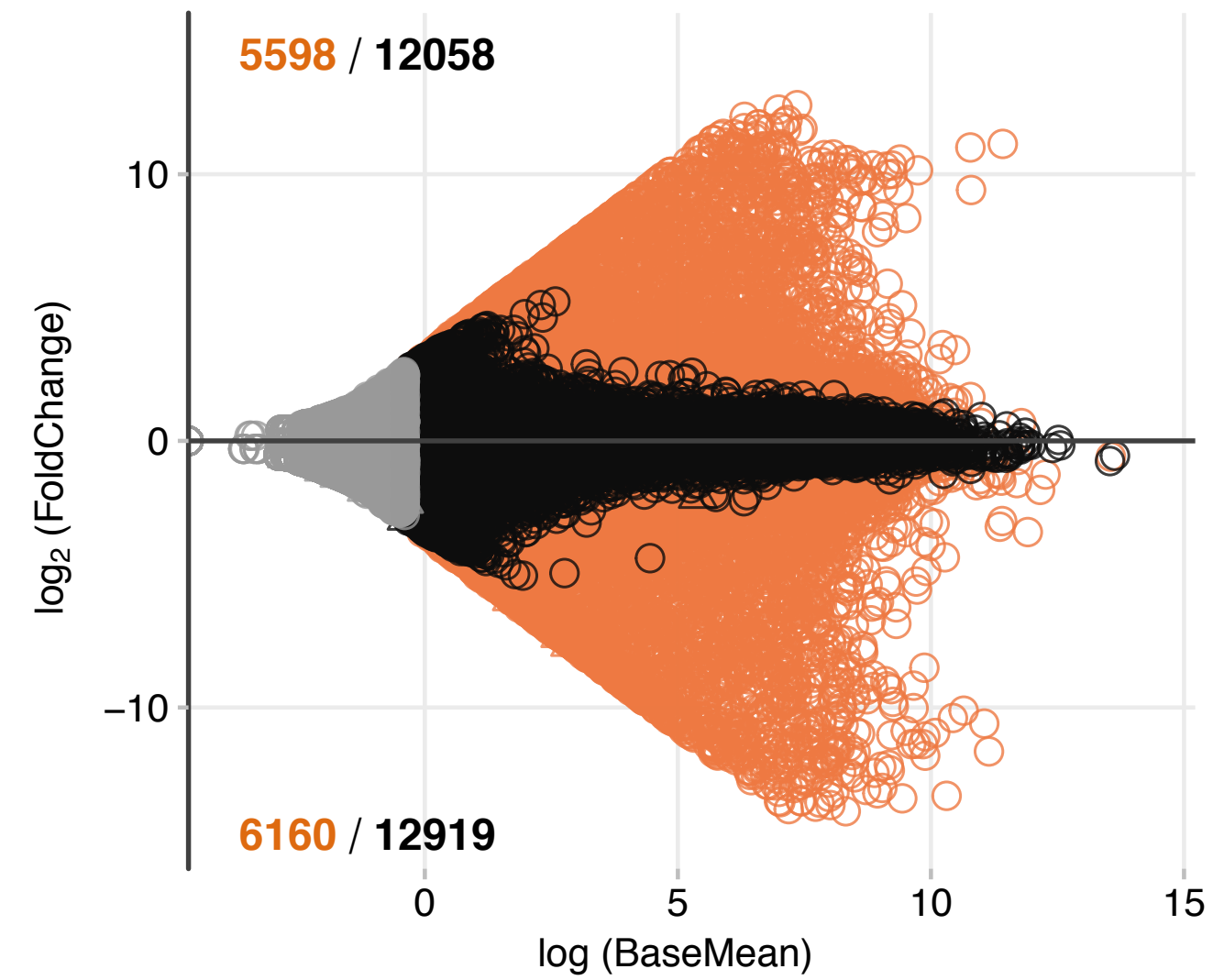
adjusted p-value:

- > 0.05
- NA
- ≤ 0.05

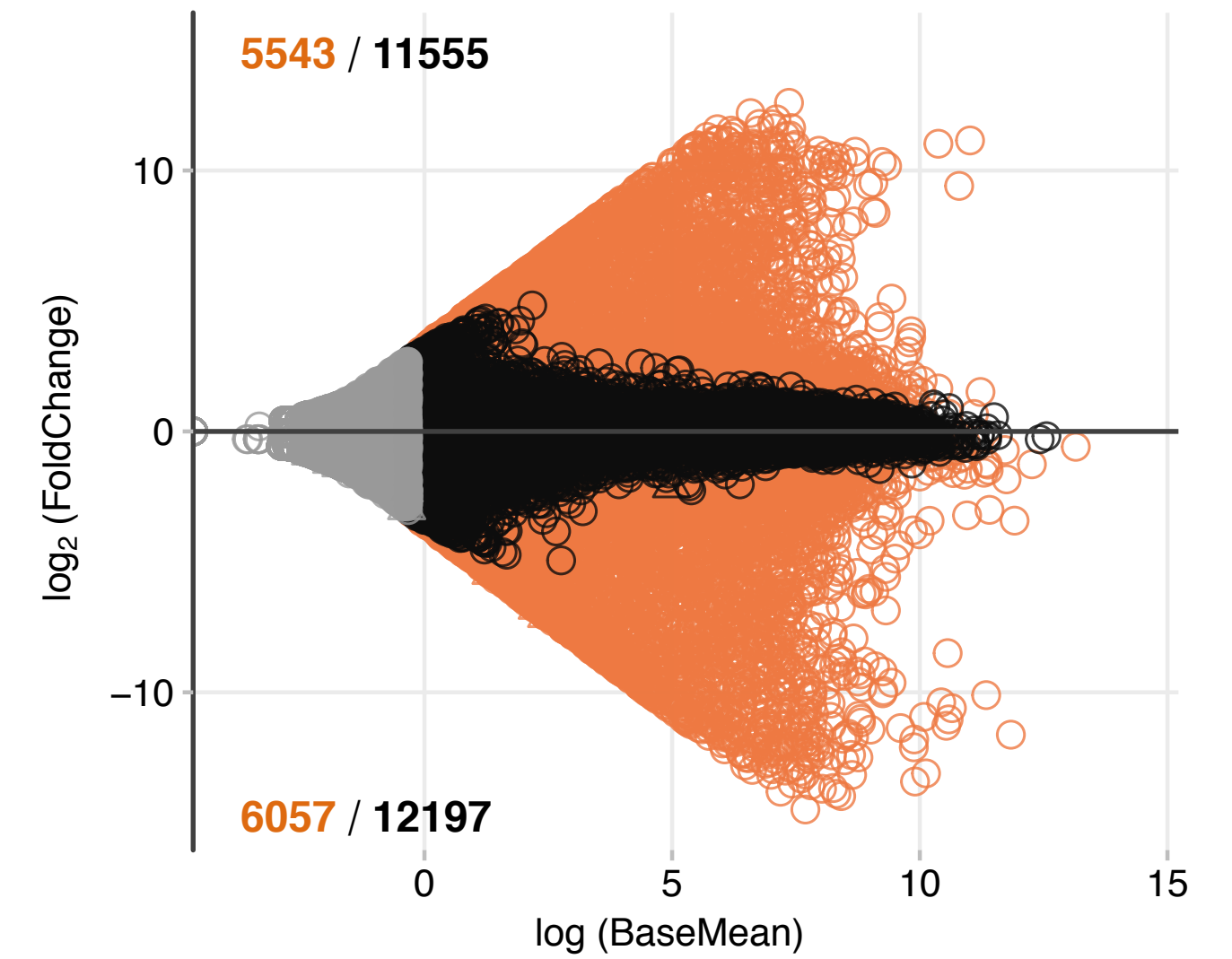
type of K562 SV, linkage, and effect:

- △ DEL__closest_enhancer_loss
- ▲ DEL__linkage_enhancer_loss
- ▽ INS__closest_enhancer_gain
- none

K562 vs GM12878, all genes MA plot



.. based on CN adjusted counts



only genes w K562 SVs in enhancer

