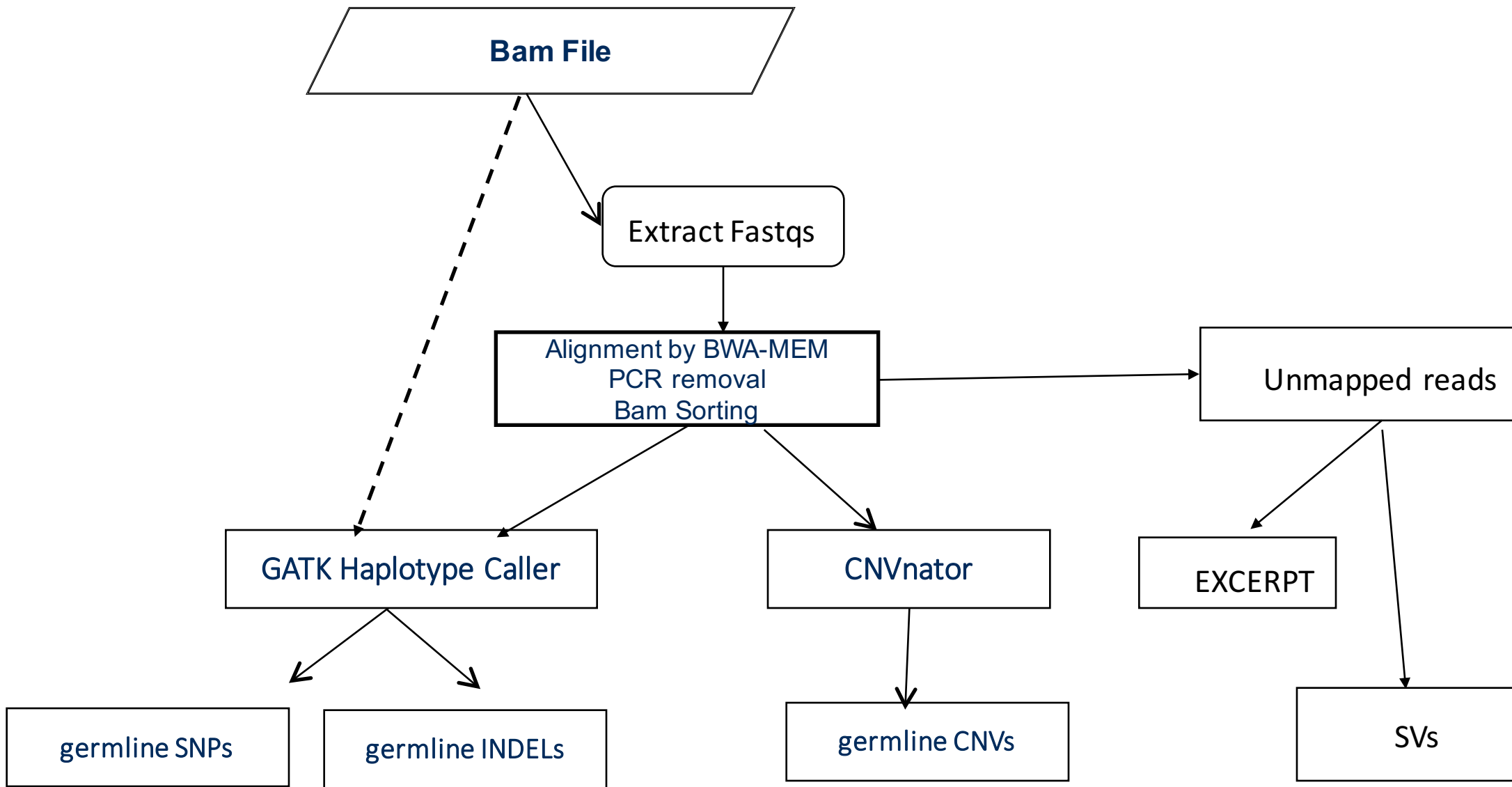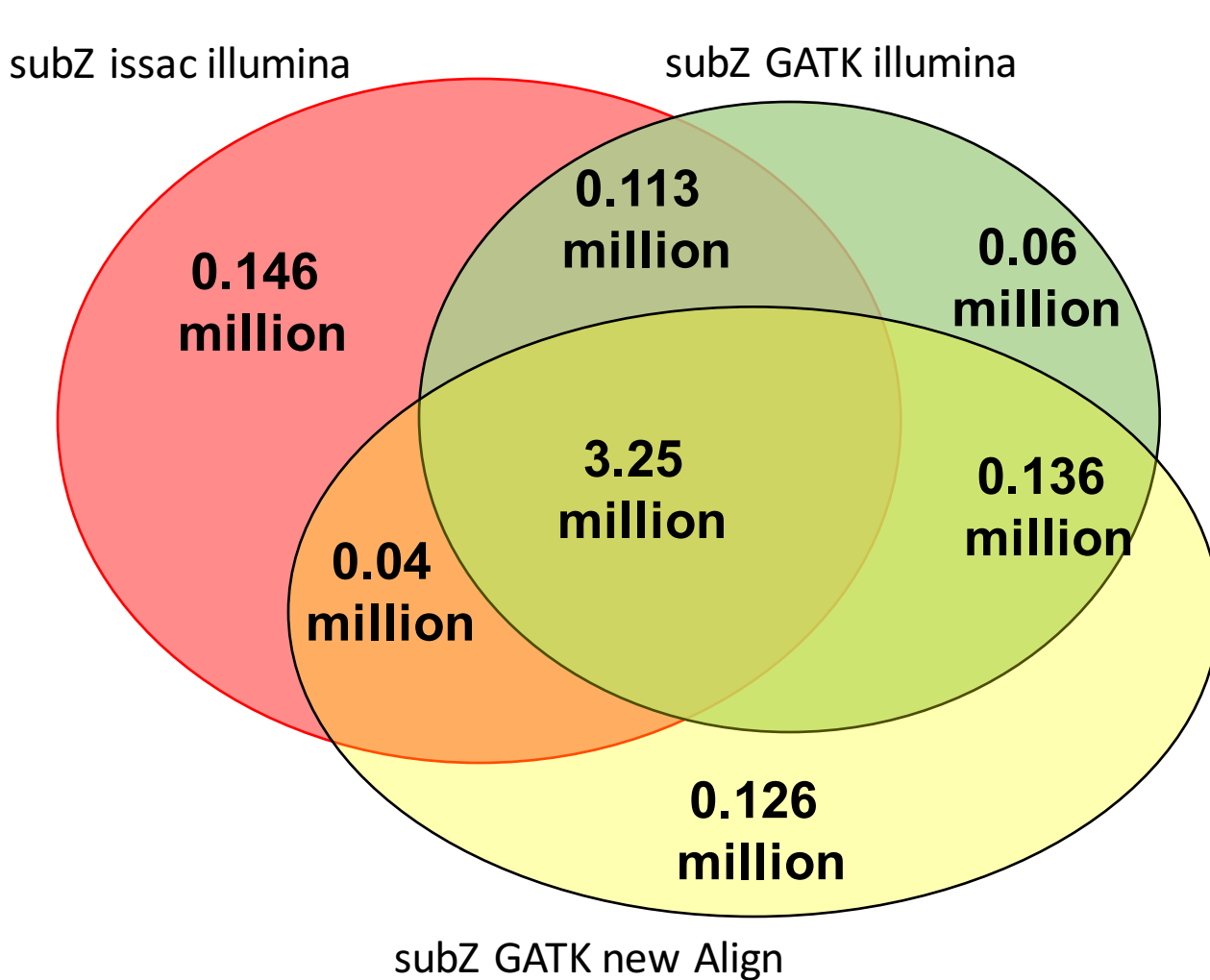# Personal Genome Analysis
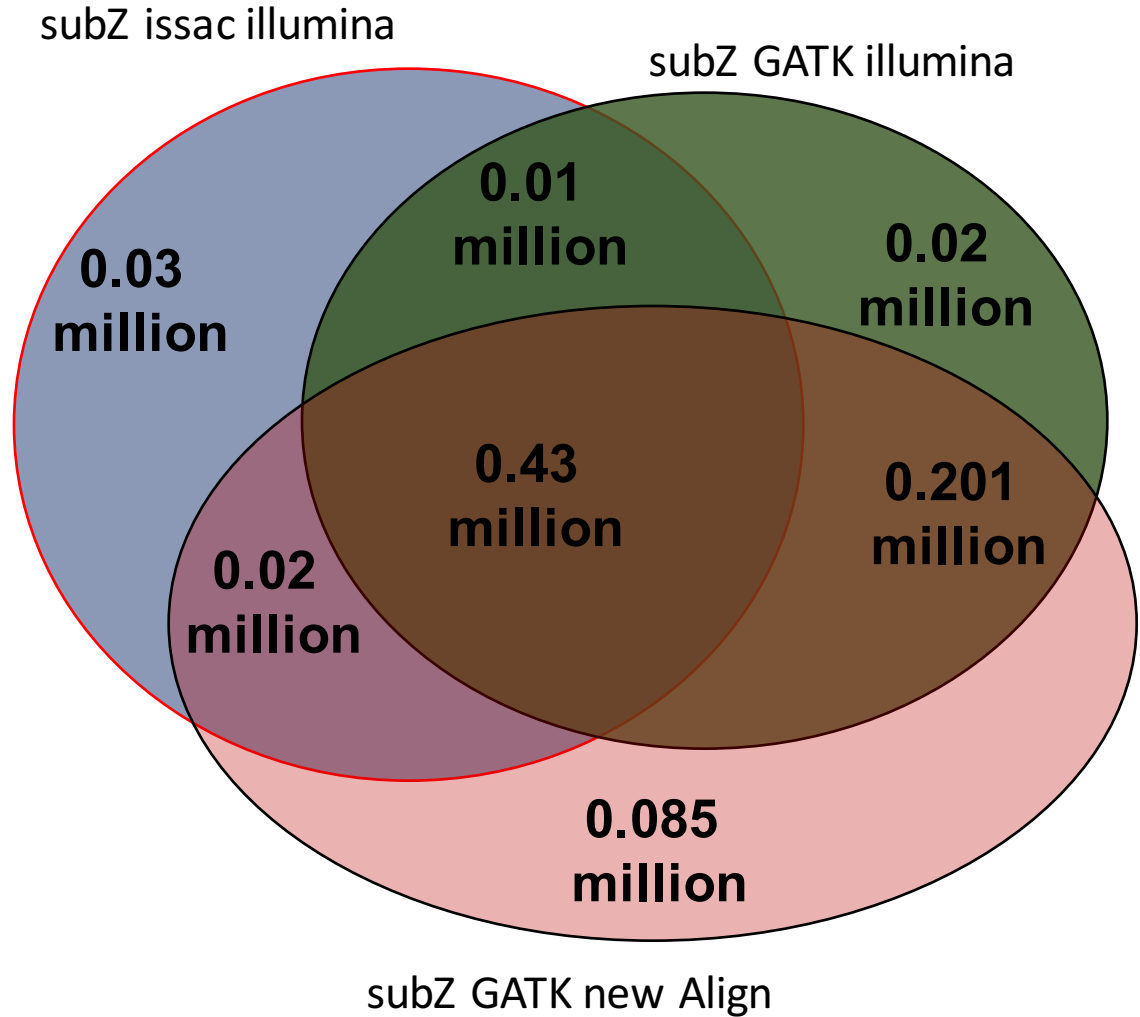
Pipeline to generate variants for personal genome analysis

Comparison of calls for subjectZ w/o post filtering

SNPs
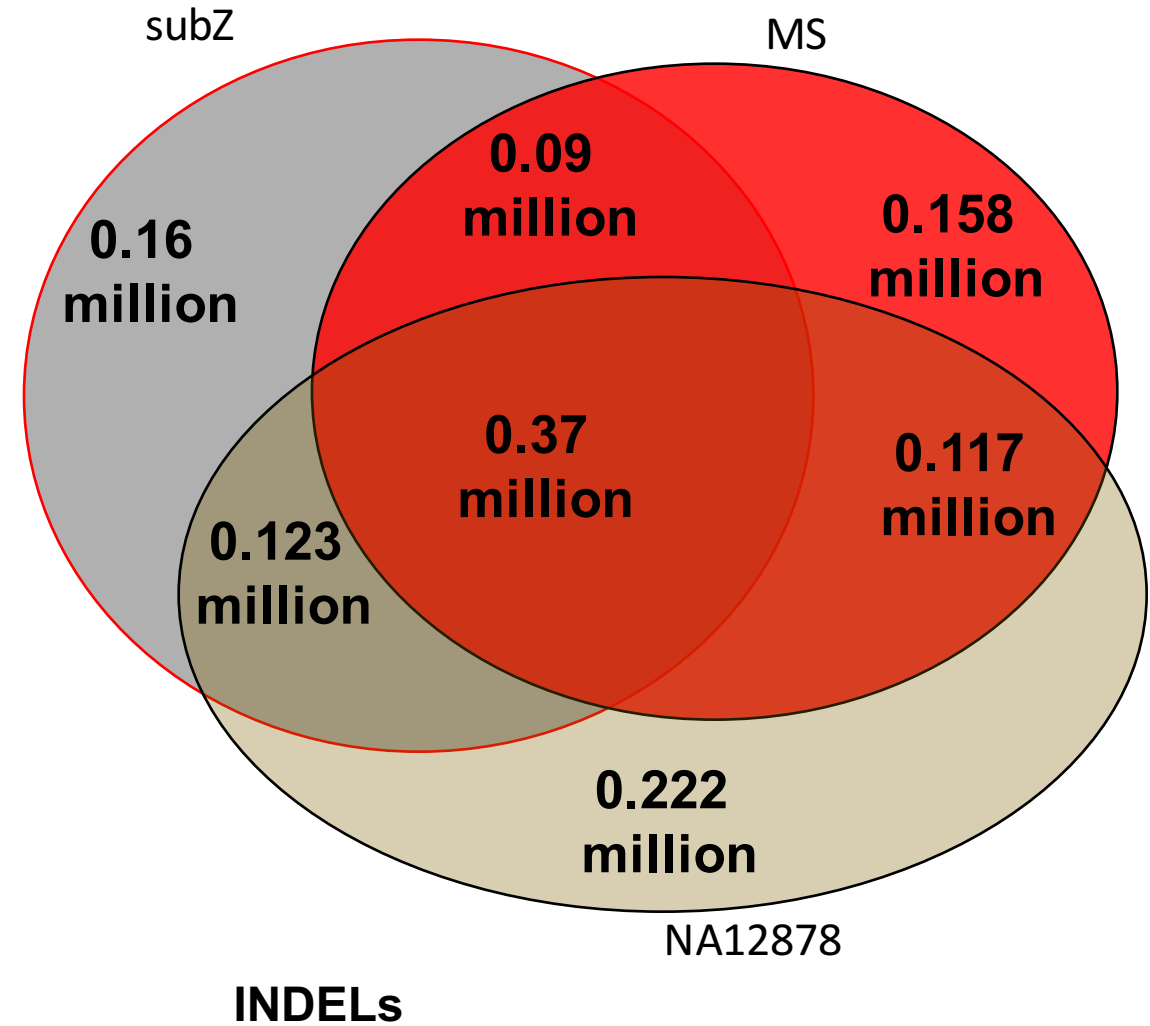
subZ issac illumina   subZ GATK illumina

0.146 million

0.113 million

0.06 million

0.04 million

3.25 million

0.136 million

0.126 million

subZ GATK new Align

INDELs

subZ issac illumina   subZ GATK illumina

0.03 million

0.01 million

0.02 million

0.02 million

0.43 million

0.201 million

0.085 million

subZ GATK new Align

Comparison of calls across three genomes w/o post filtering

SNPs

- subZ
- MS
- 0.816 million
- 0.504 million
- 0.78 million
- 1.73 million
- 0.504 million
- 0.506 million
- 0.85 million
- NA12878

INDELs

- subZ
- MS
- 0.16 million
- 0.09 million
- 0.158 million
- 0.37 million
- 0.117 million
- 0.123 million
- 0.222 million
- NA12878
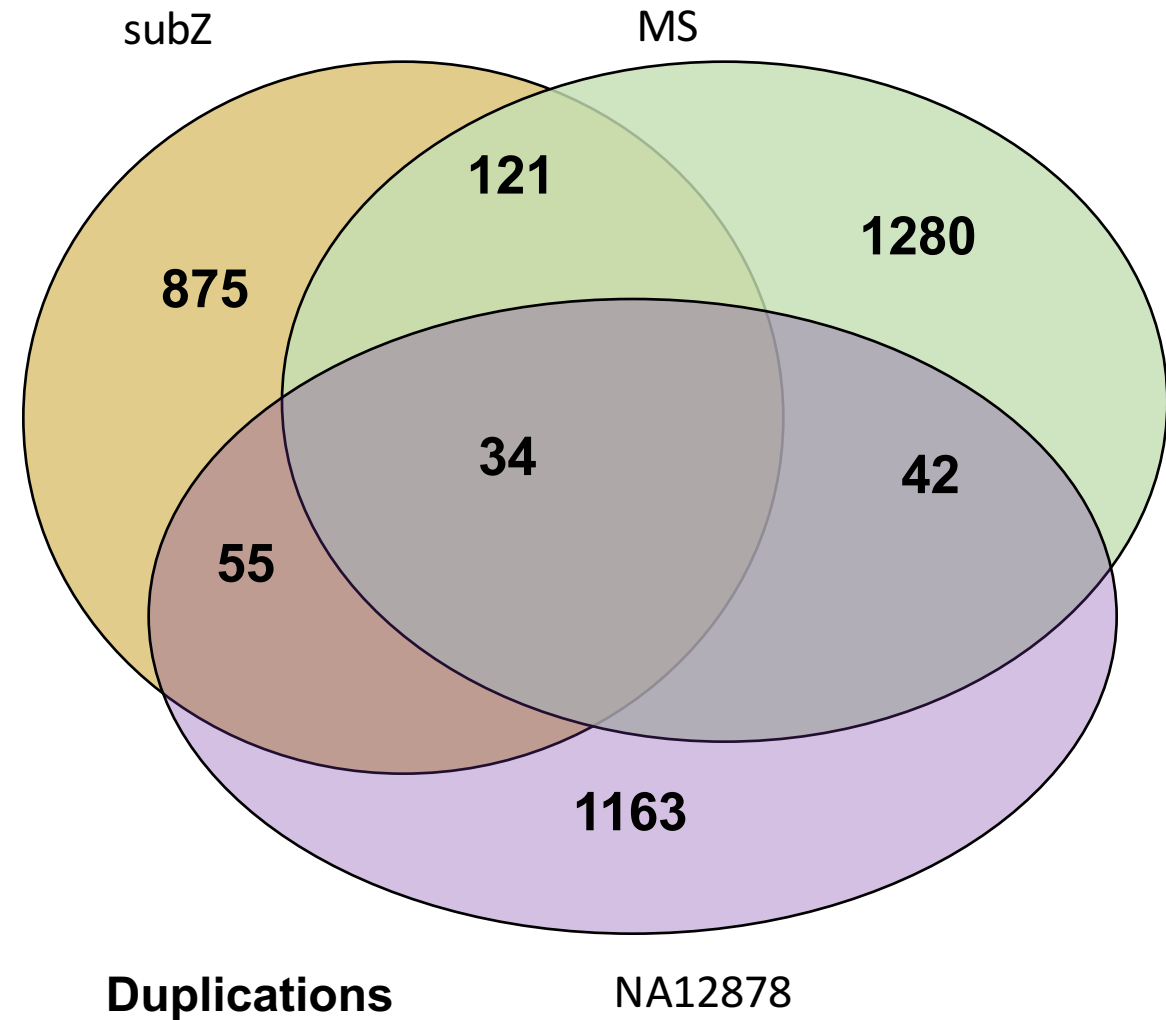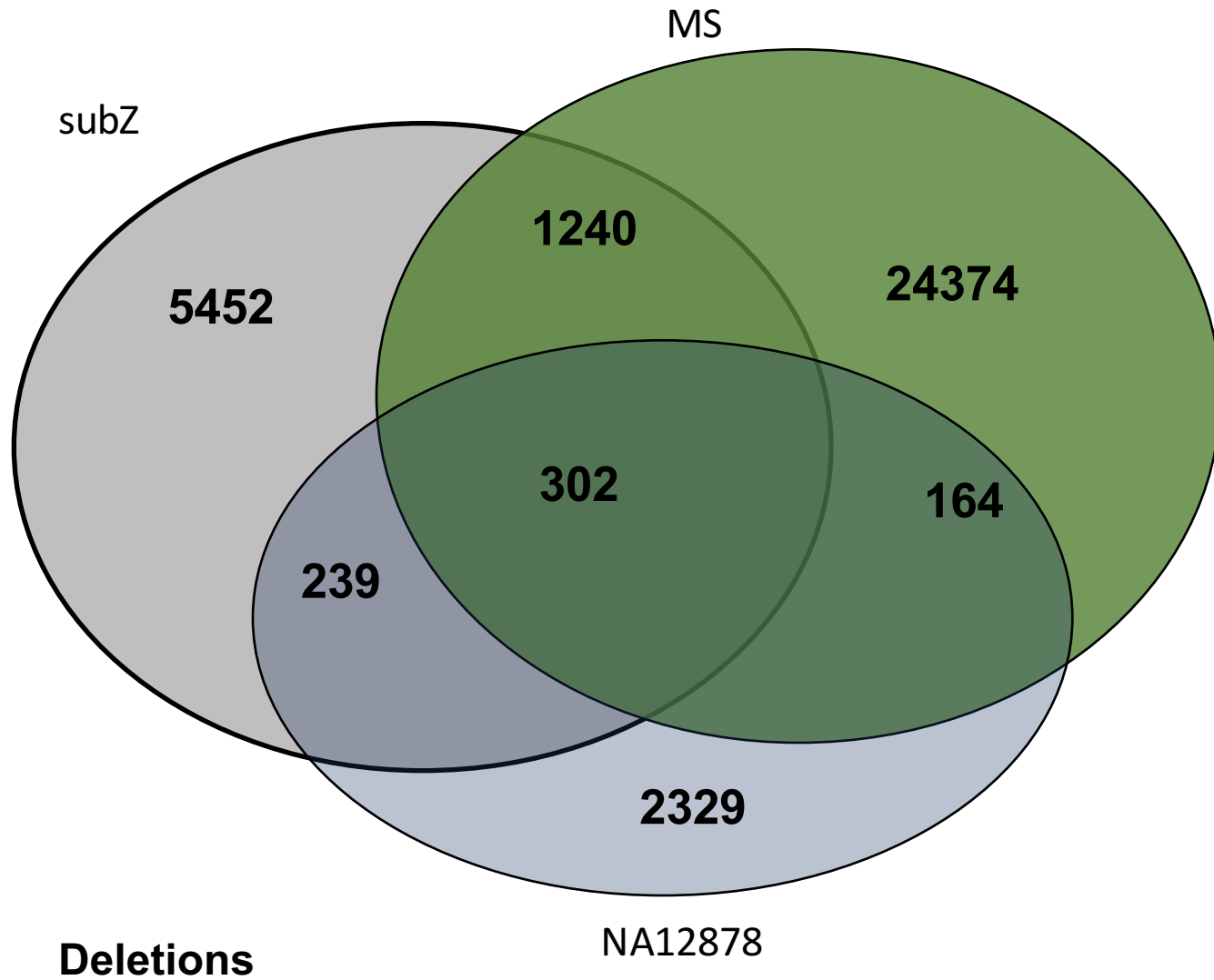
Comparison of calls across three genomes w/o post filtering

Deletions — subZ, MS, NA12878:
- subZ only: 5452
- subZ ∩ MS: 1240
- MS only: 24374
- subZ ∩ MS ∩ NA12878: 302
- MS ∩ NA12878: 164
- subZ ∩ NA12878: 239
- NA12878 only: 2329

Duplications — subZ, MS, NA12878:
- subZ only: 875
- subZ ∩ MS: 121
- MS only: 1280
- subZ ∩ MS ∩ NA12878: 34
- MS ∩ NA12878: 42
- subZ ∩ NA12878: 55
- NA12878 only: 1163

Comparison of calls across three genomes with post filtering

Deletions >= 2kb
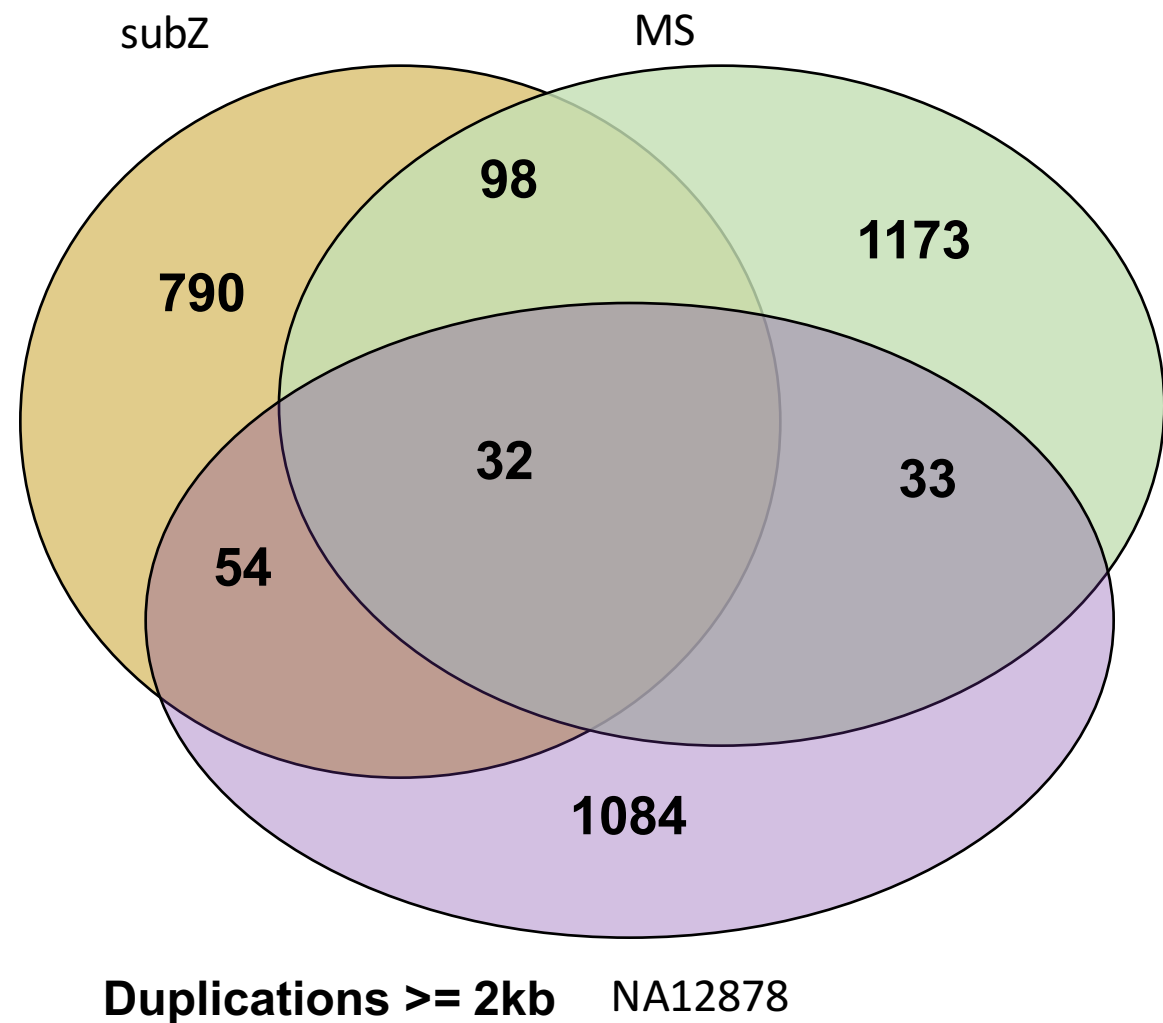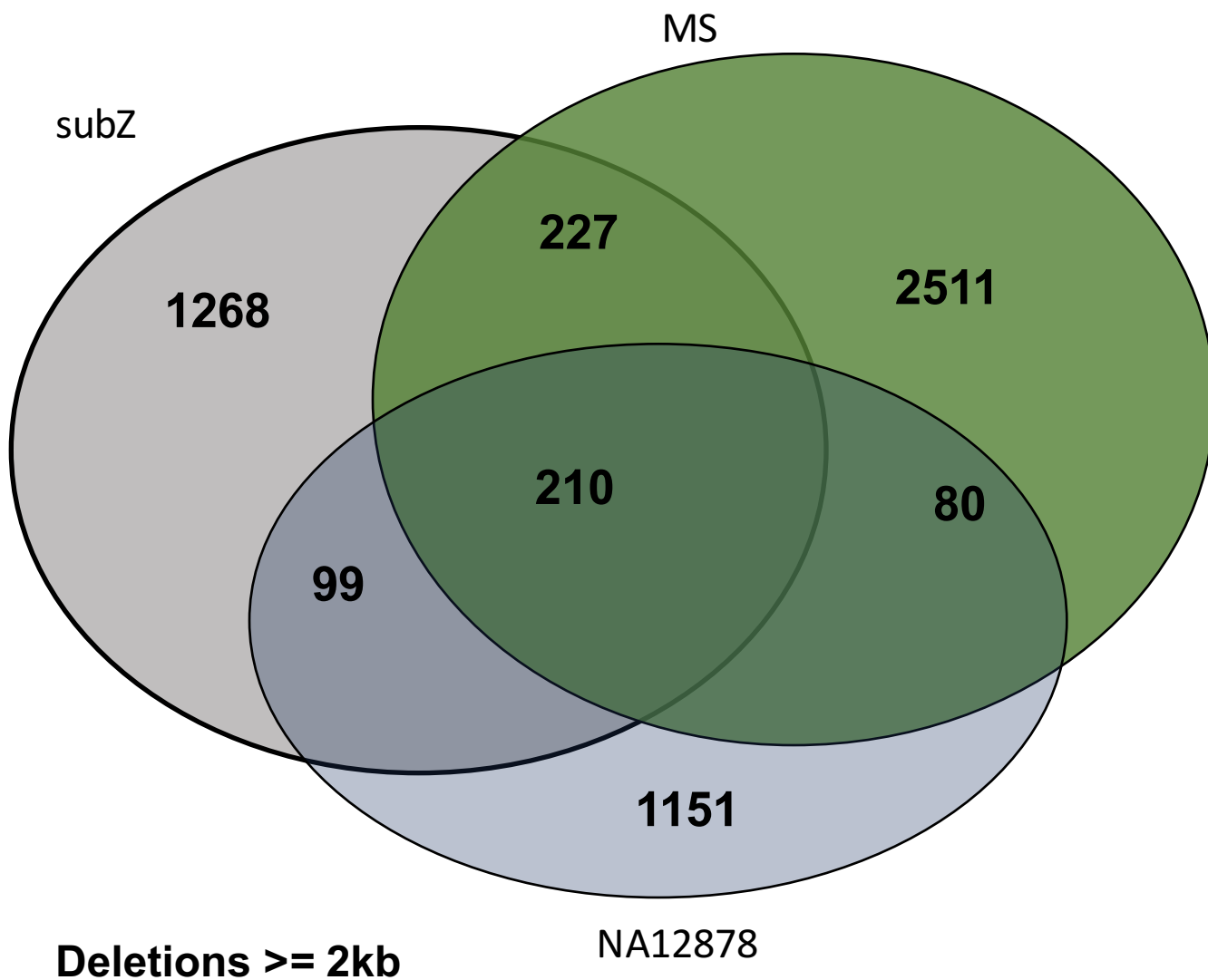
Duplications >= 2kb
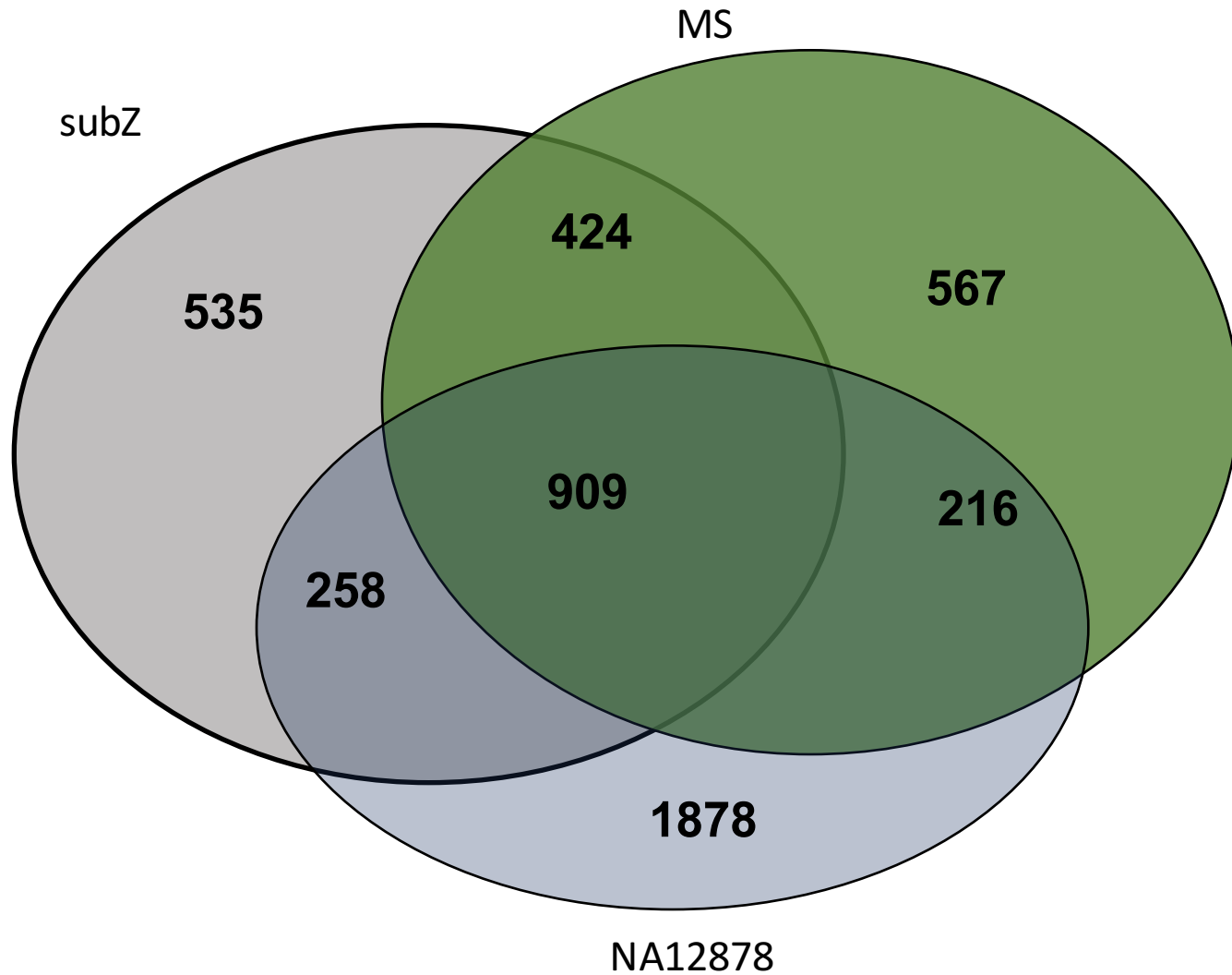
Comparison of calls across three genomes CREST calls
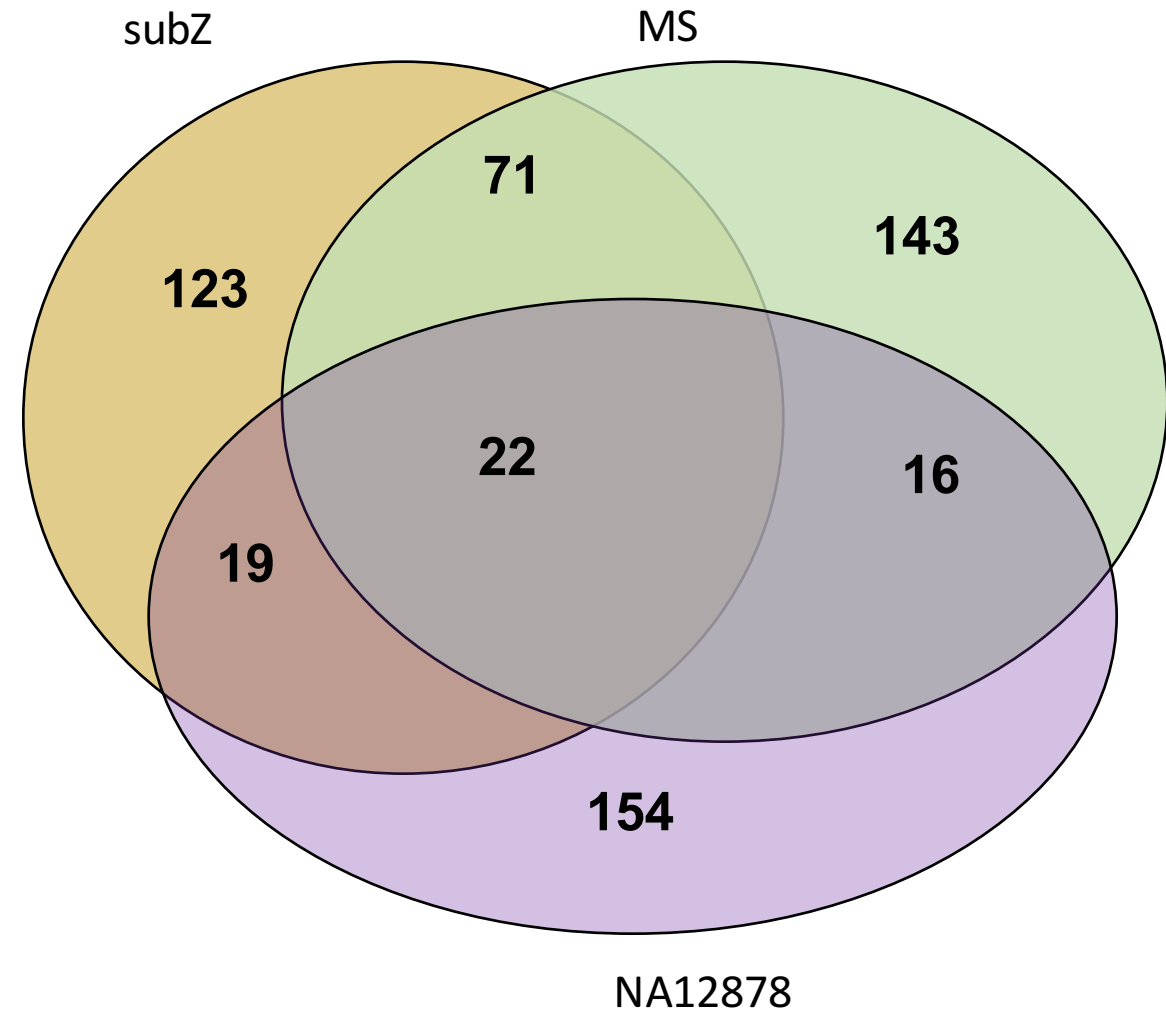
Comparison of calls across three genomes CNVnator calls consistent with CREST calls

Deletions

Duplications