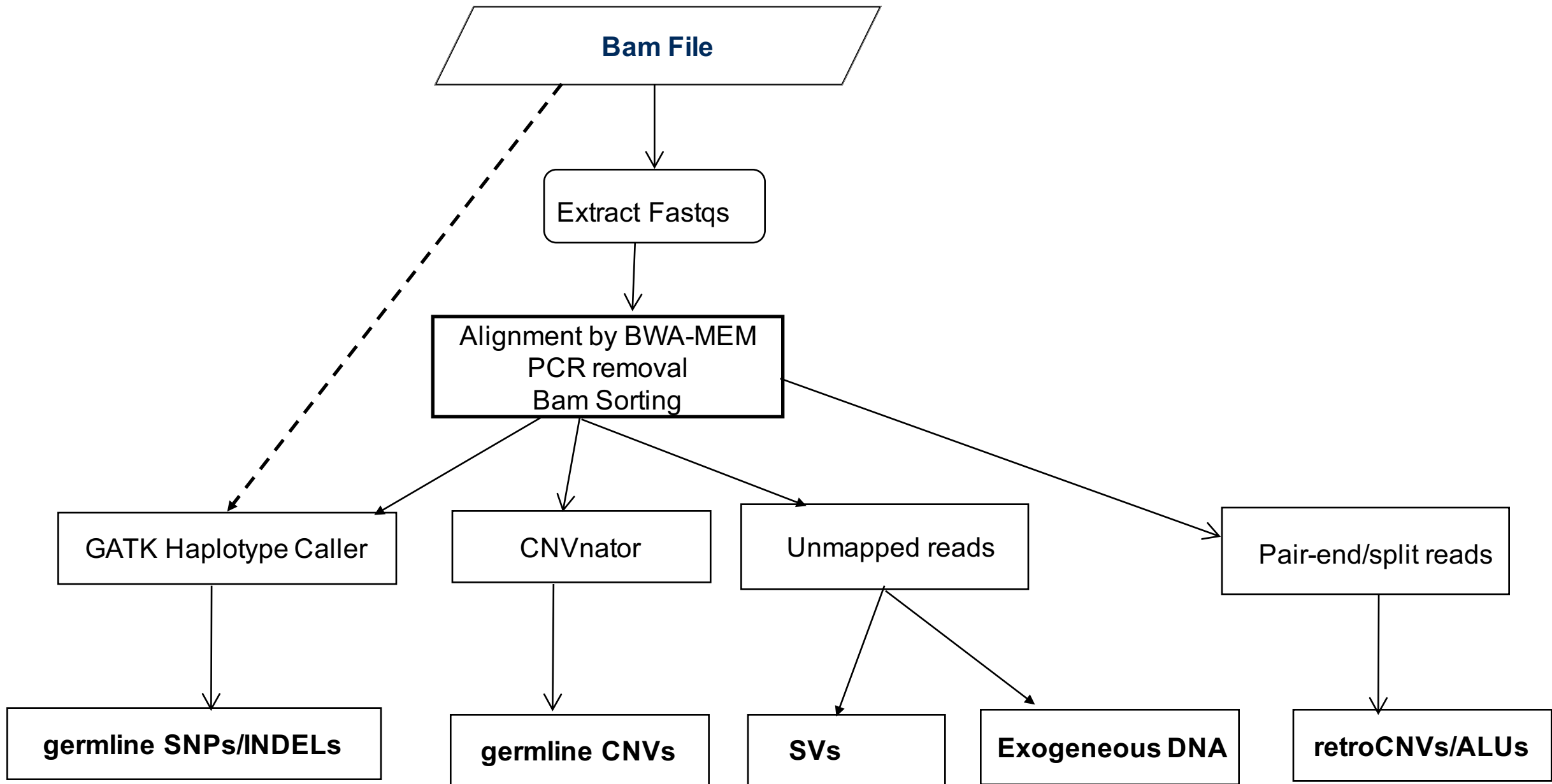
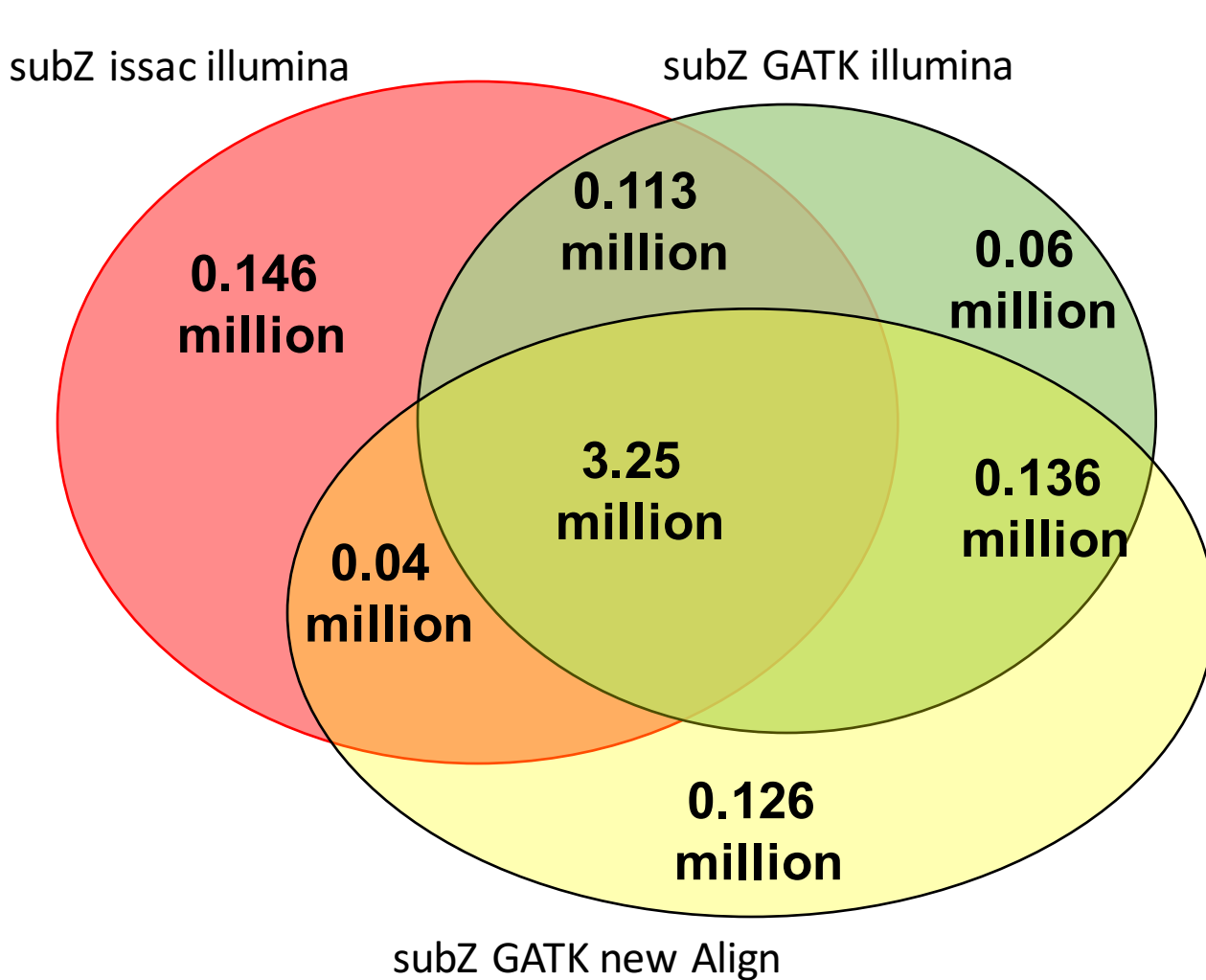


Personal Genome Analysis

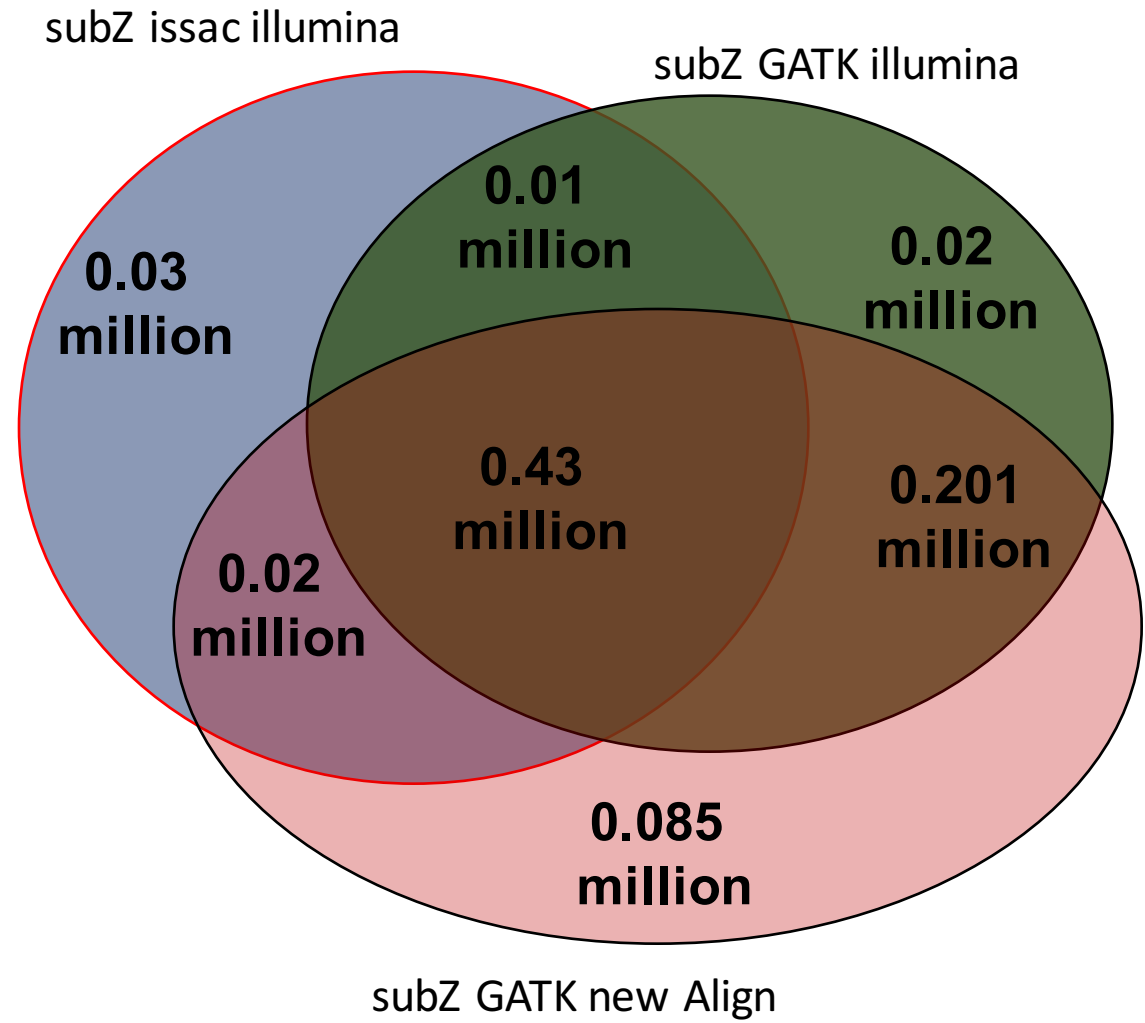


Pipeline to generate variants for personal genome analysis

Comparison of variant calls for subject Z

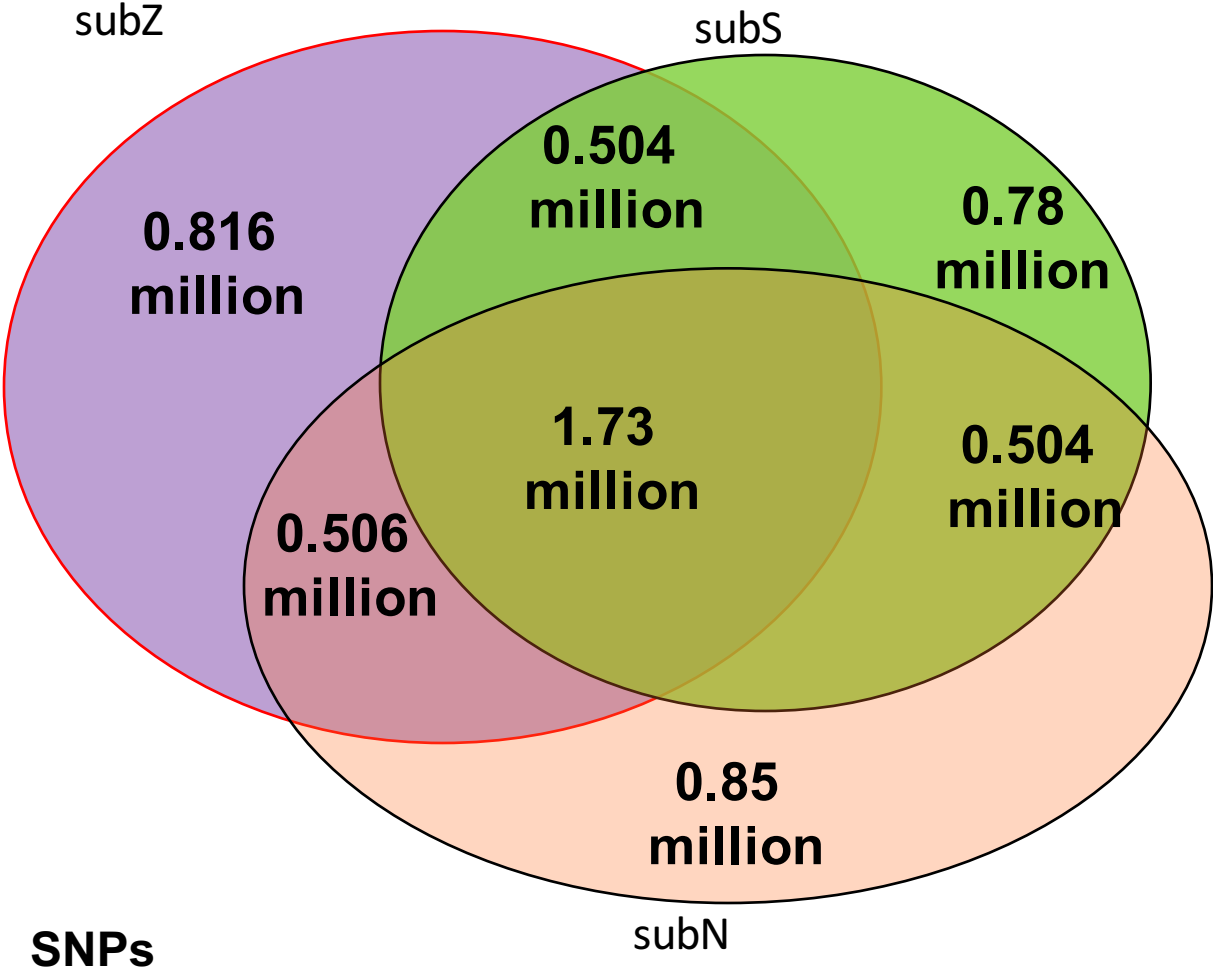


SNPs

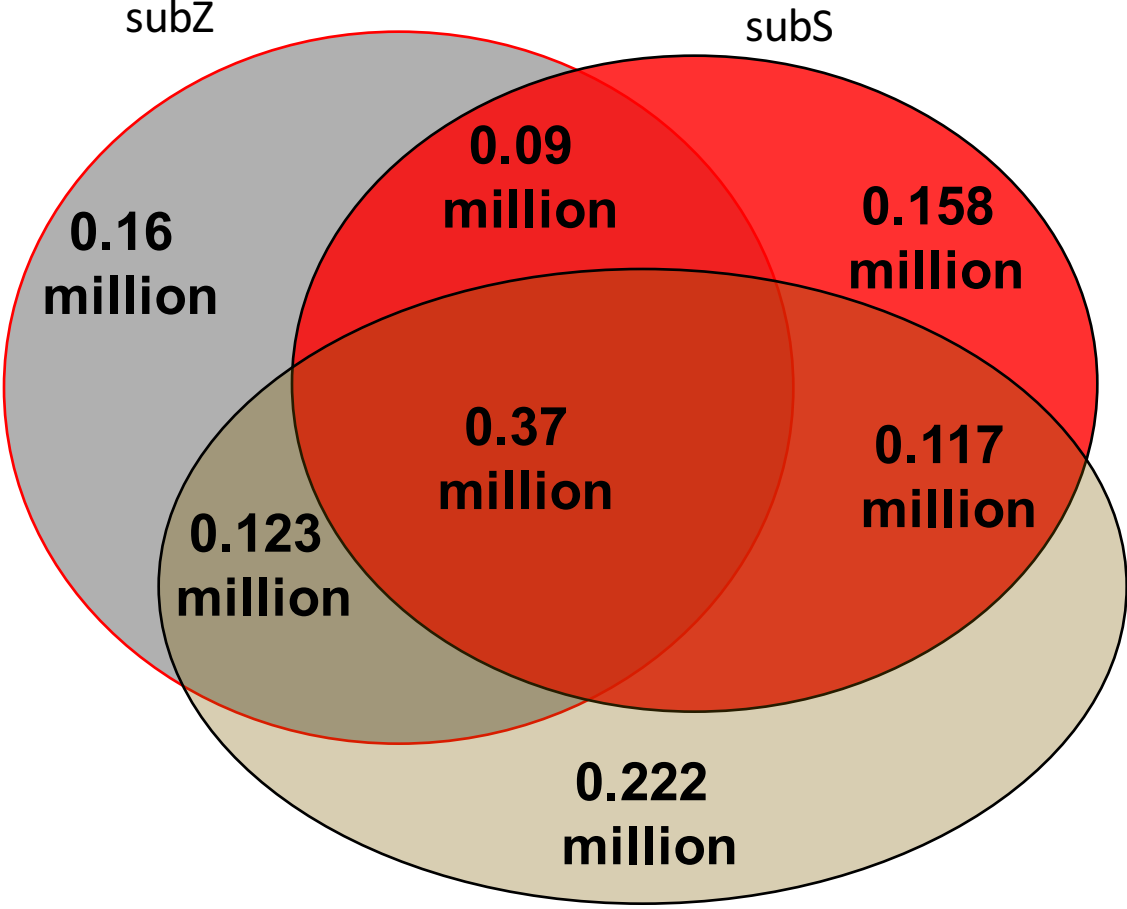


INDELs

Comparison of SNVs & INDELs across three genomes

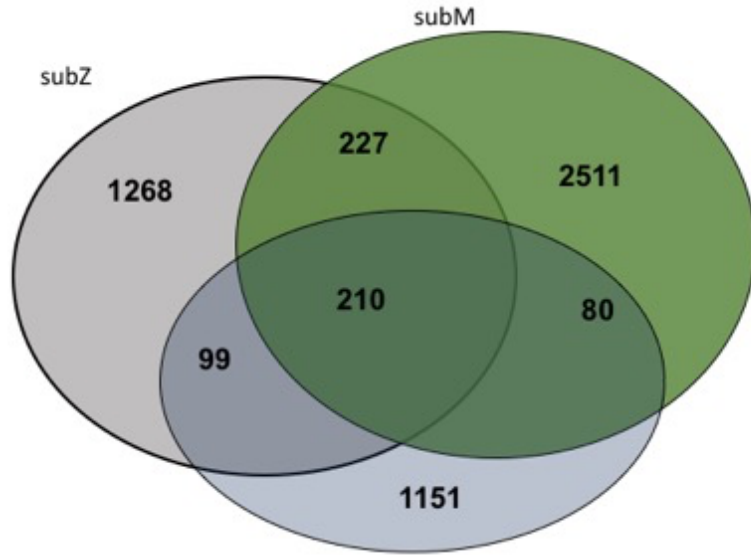


SNPs

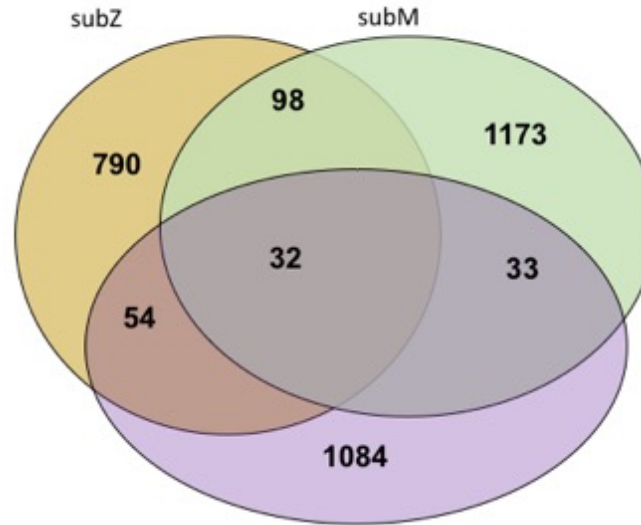


INDELs

Challenges associated with identifying structural variations



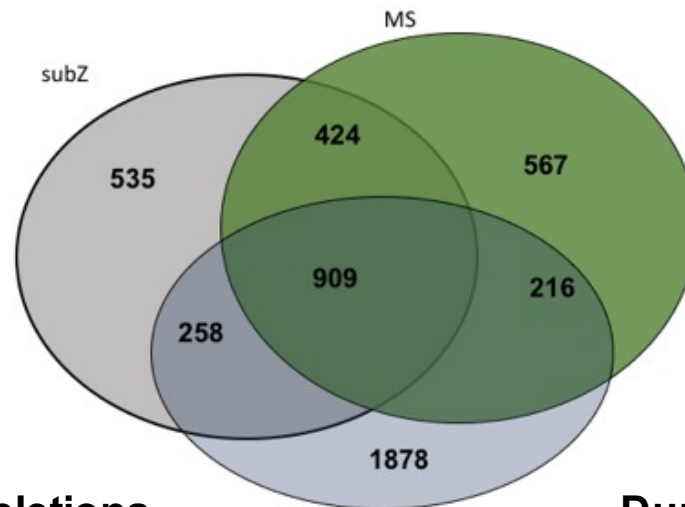
Deletions



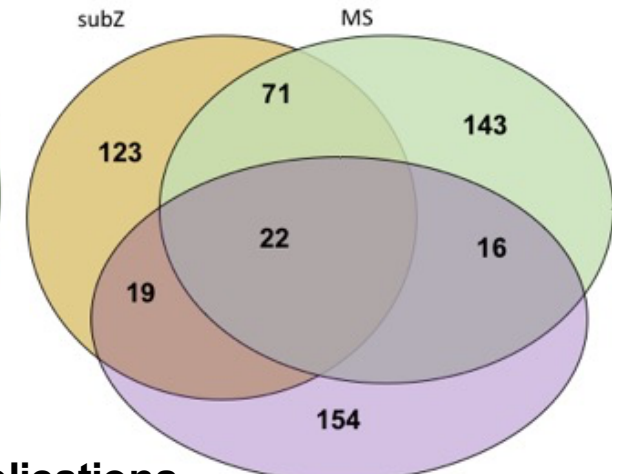
Duplications

CNVnator assigned large deletions and duplications

CREST assigned large deletions and duplications



Deletions



Duplications

NA12878

NA12878

Consensus structural variations across three genomes

