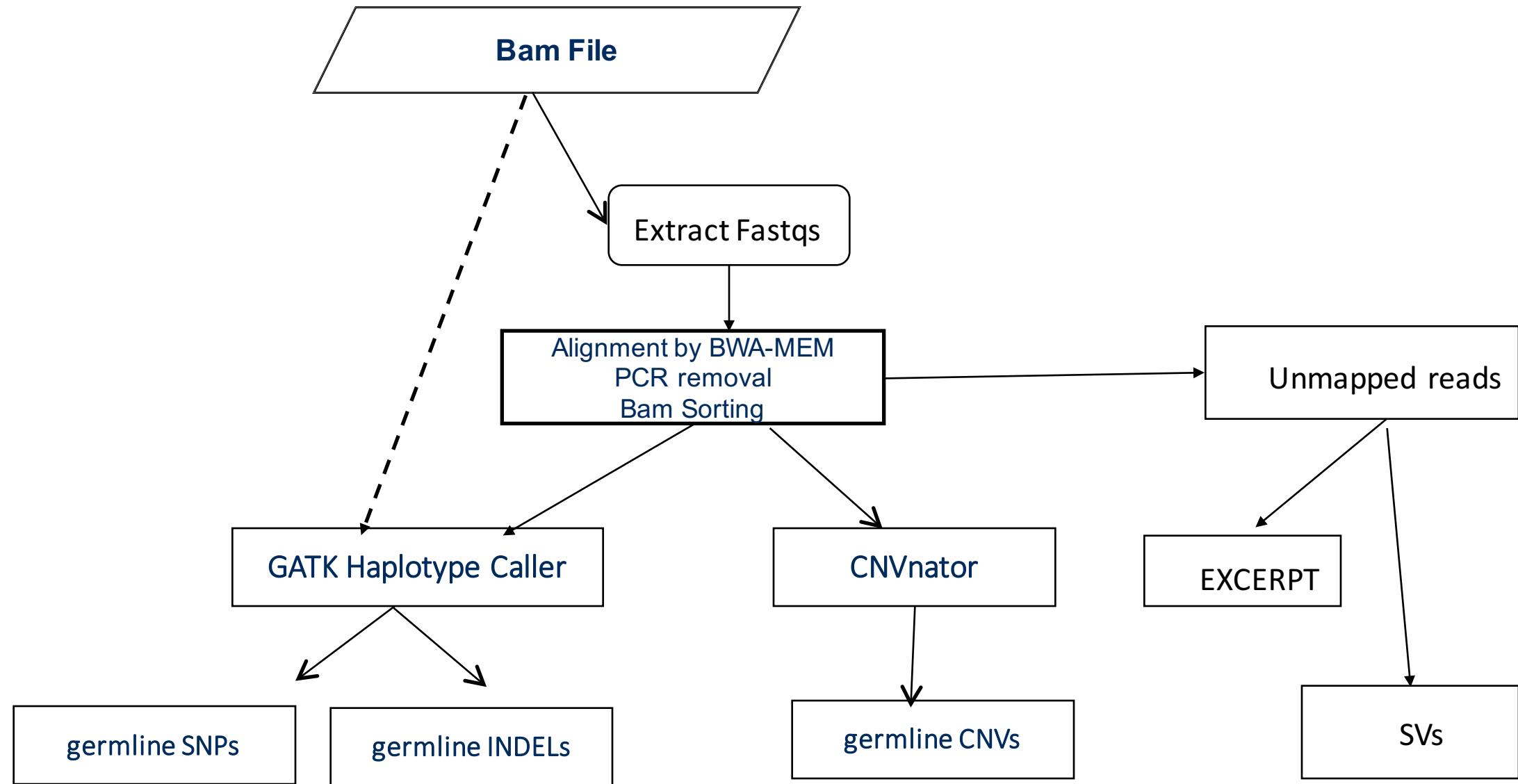


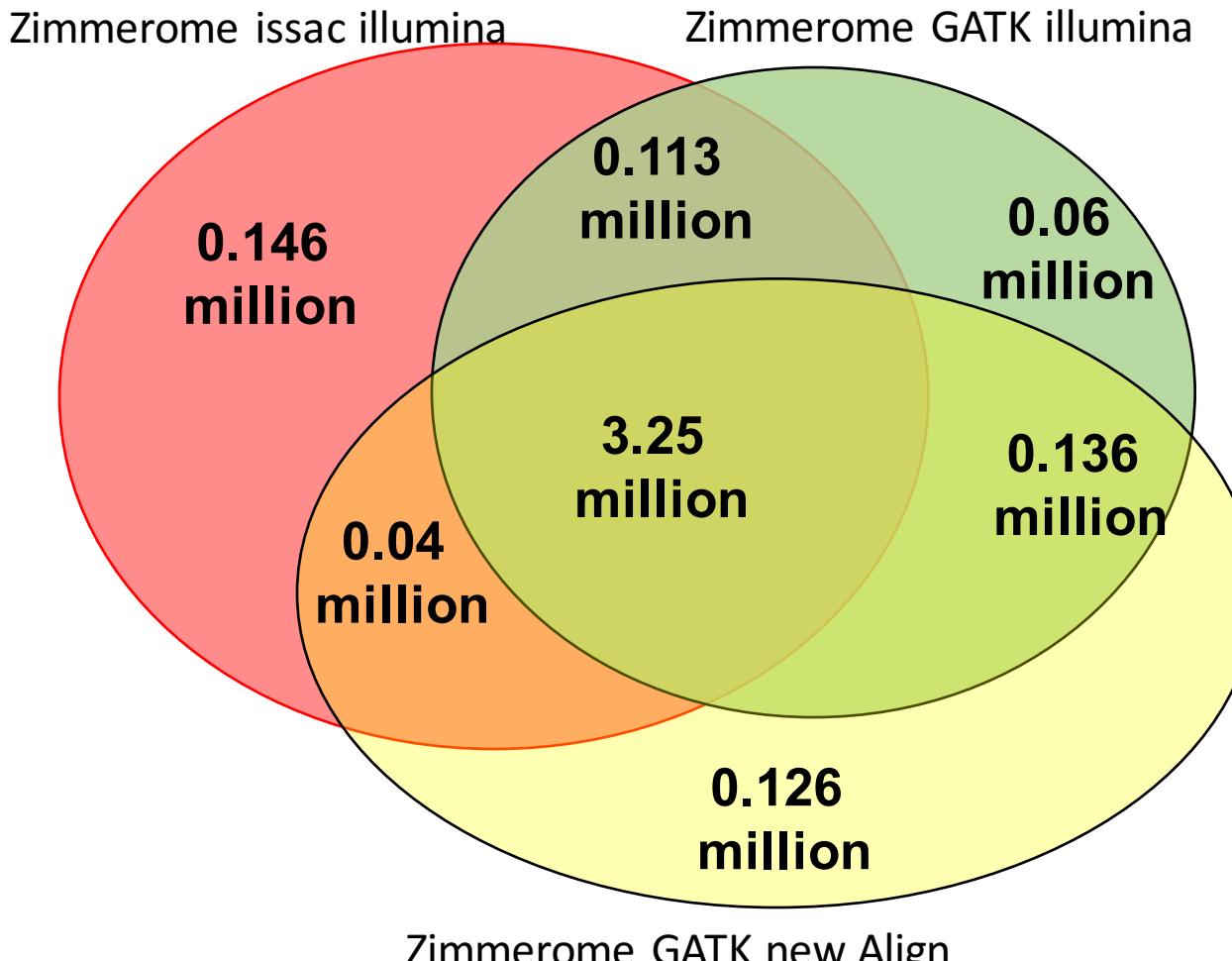
Zimmerome Analysis

P2-tech

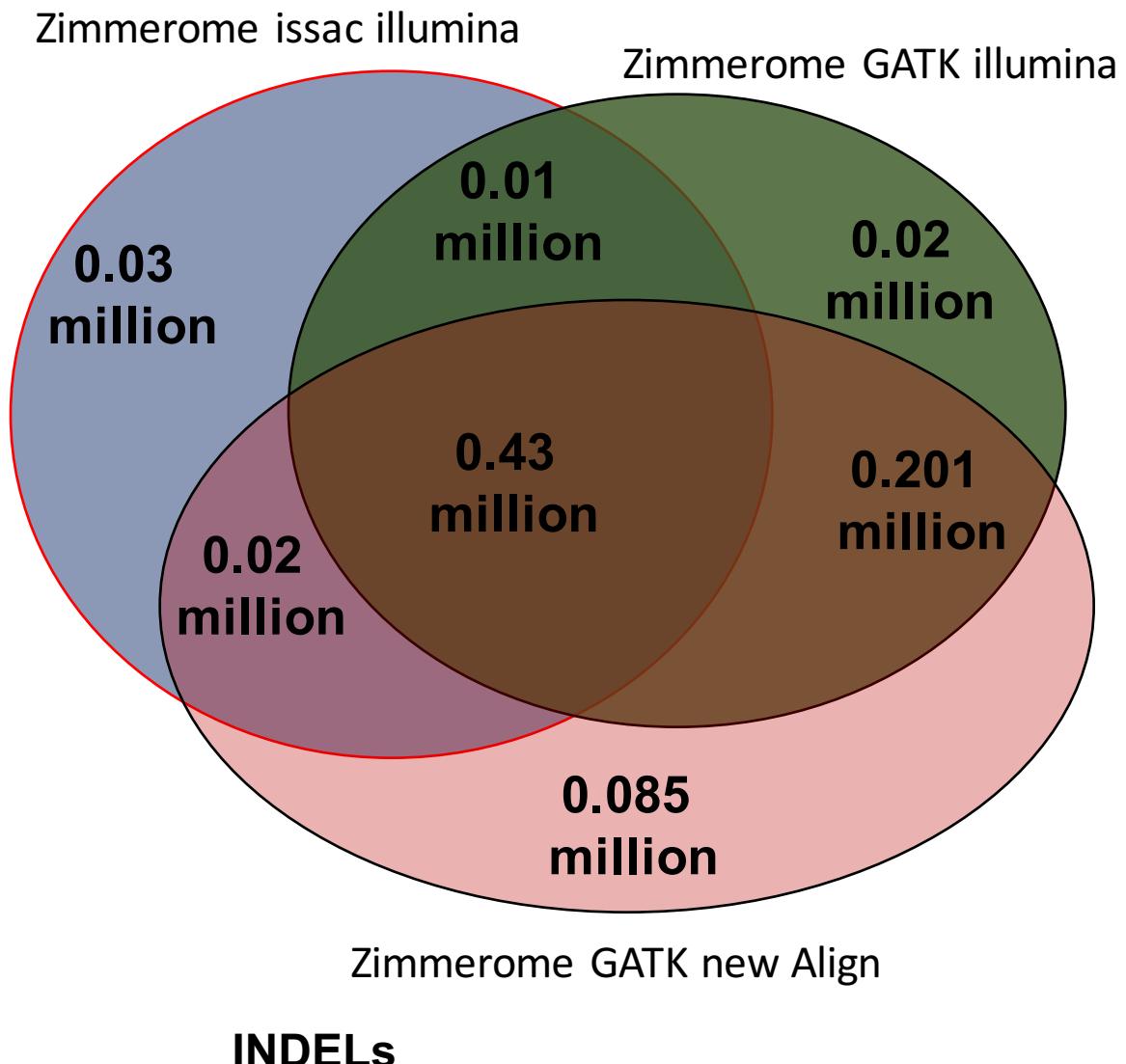


Pipeline to generate variants for personal genome analysis

Comparison of calls for zimmerome w/o post filtering

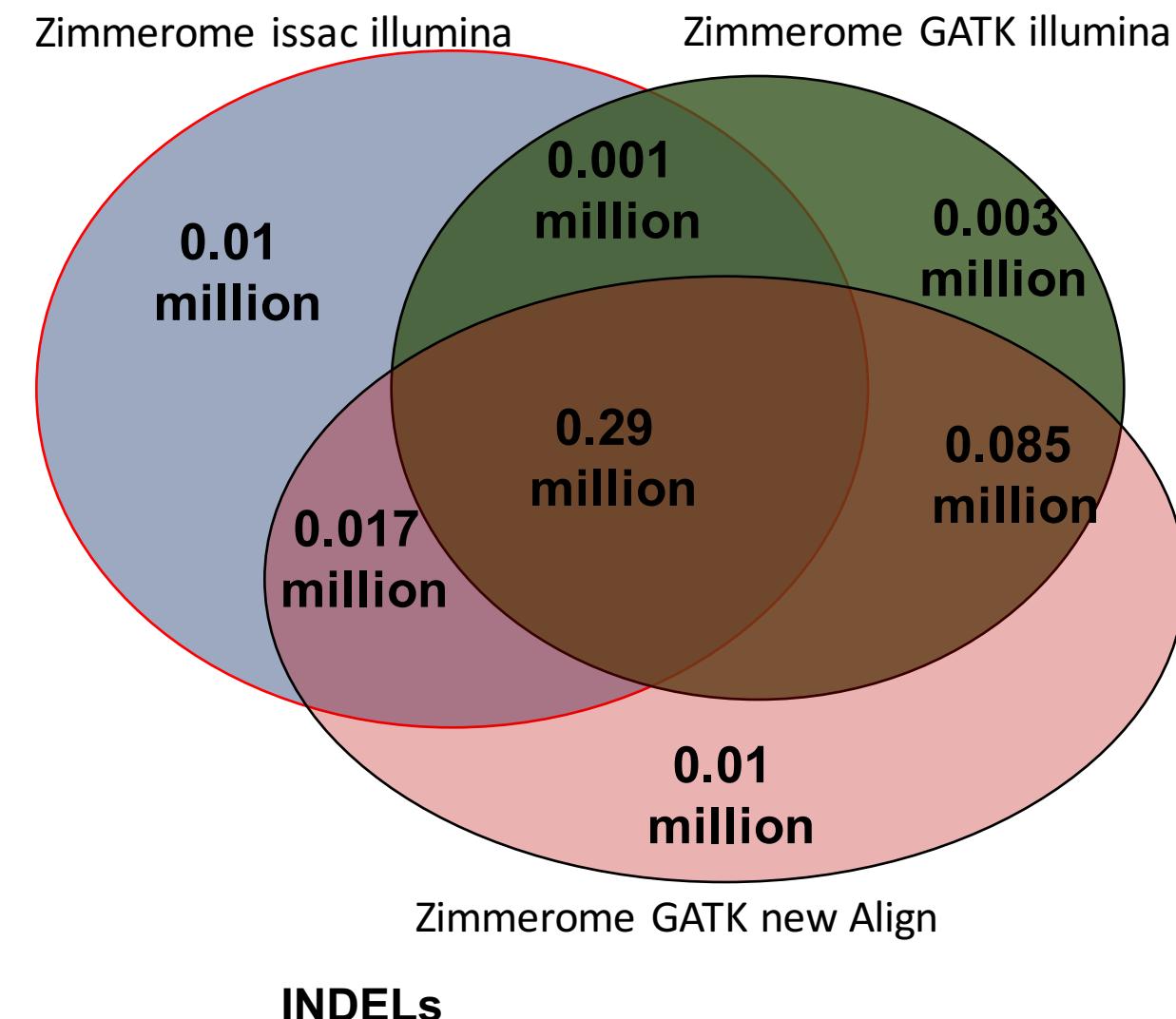
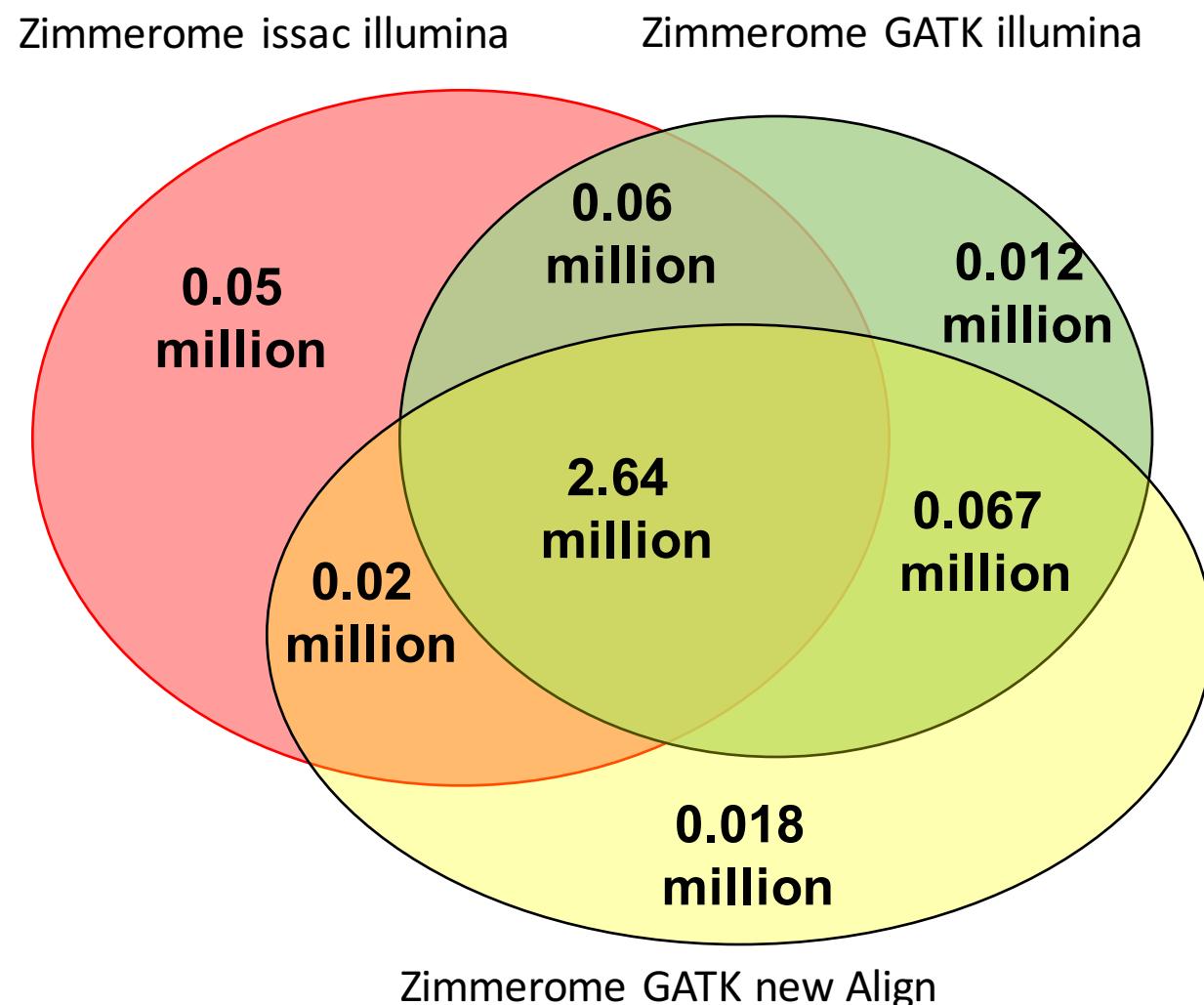


SNPs

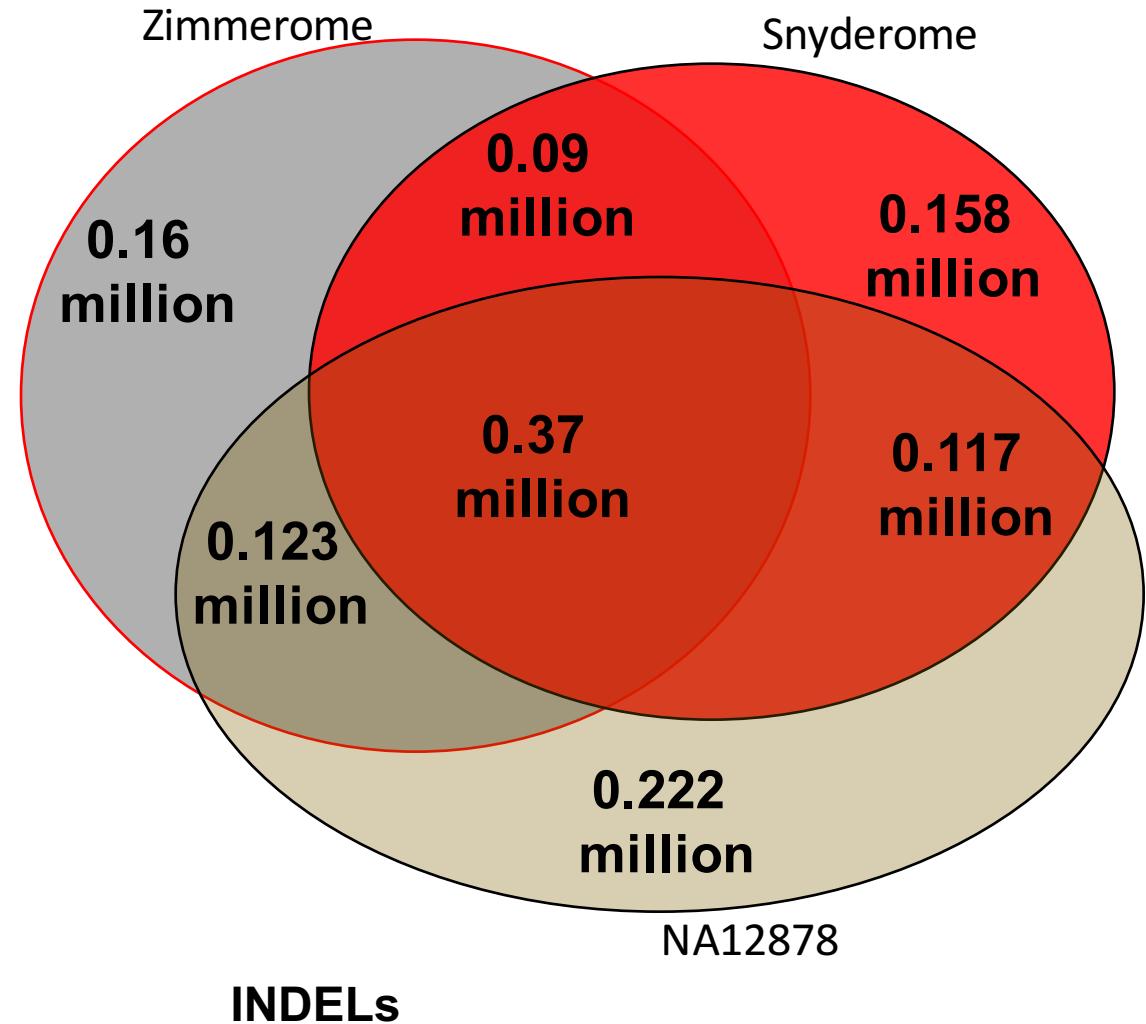
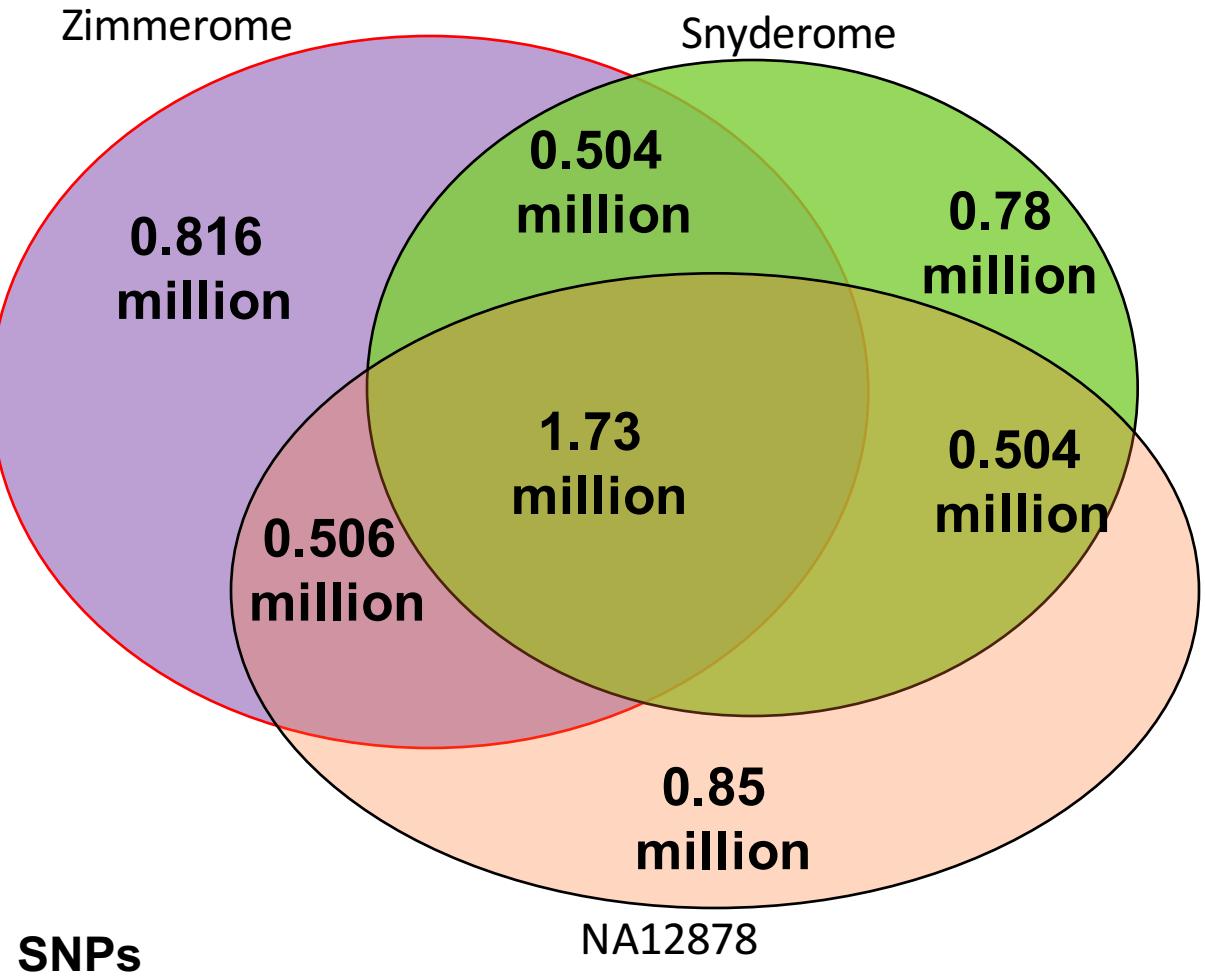


INDELs

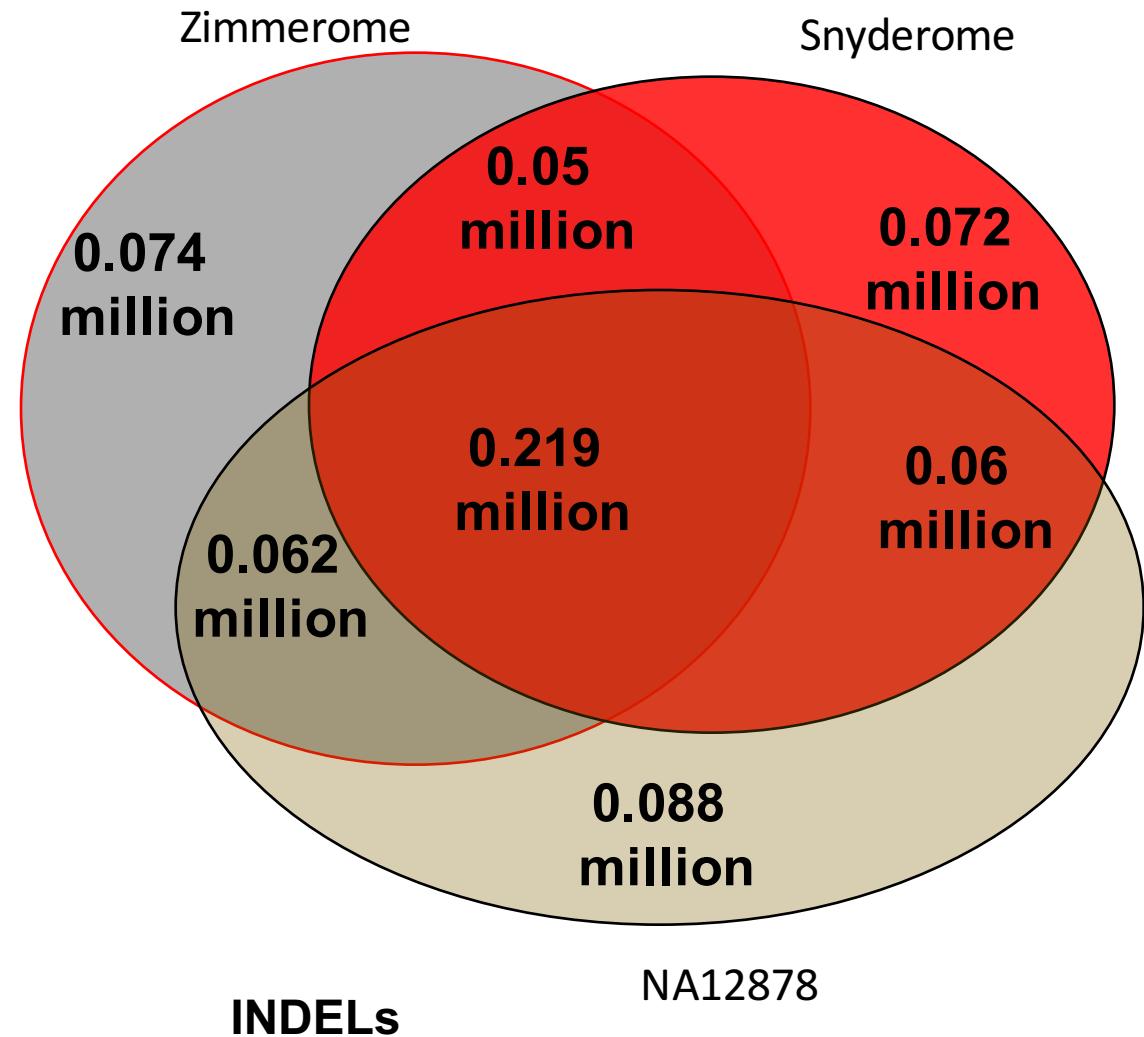
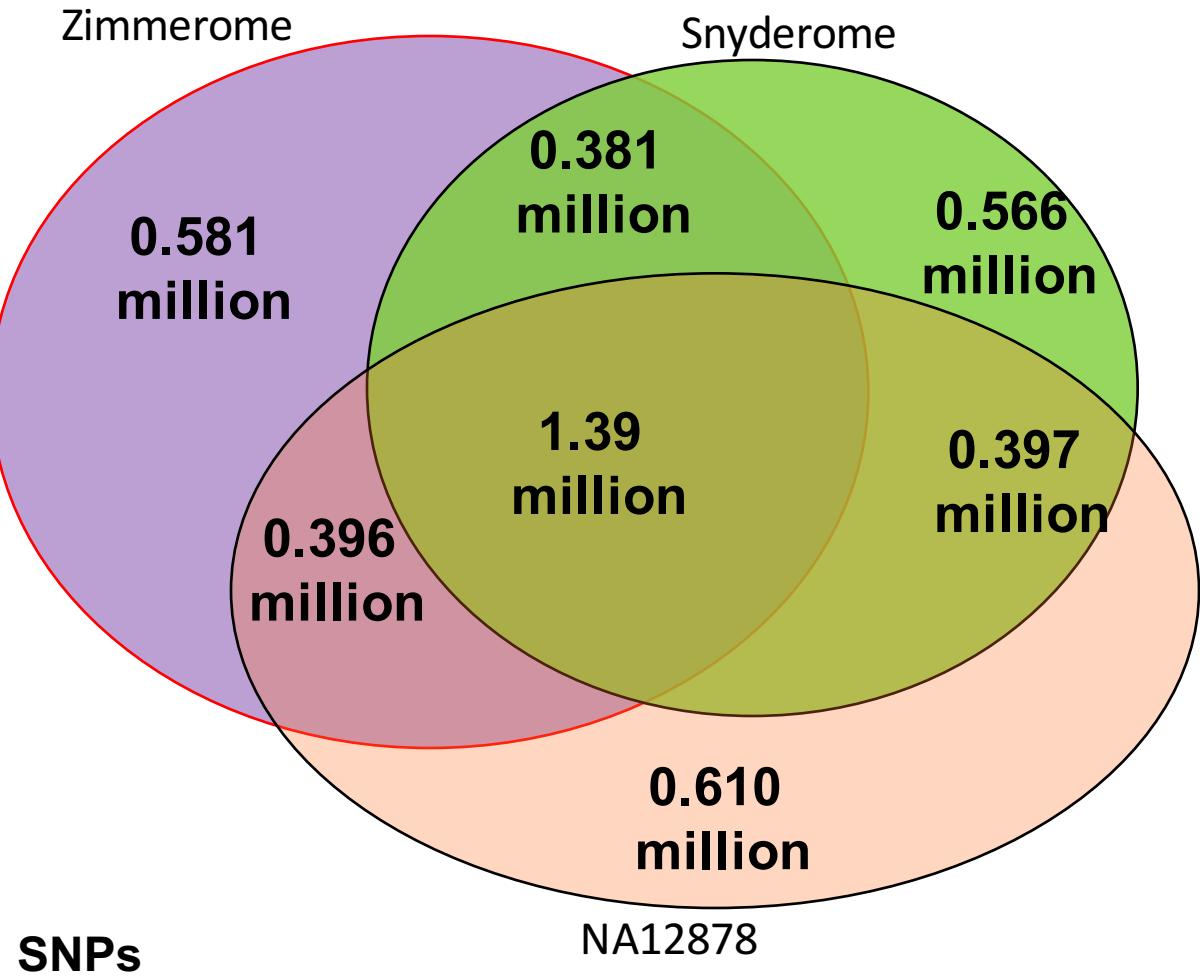
Comparison of calls for zimmerome after filtering



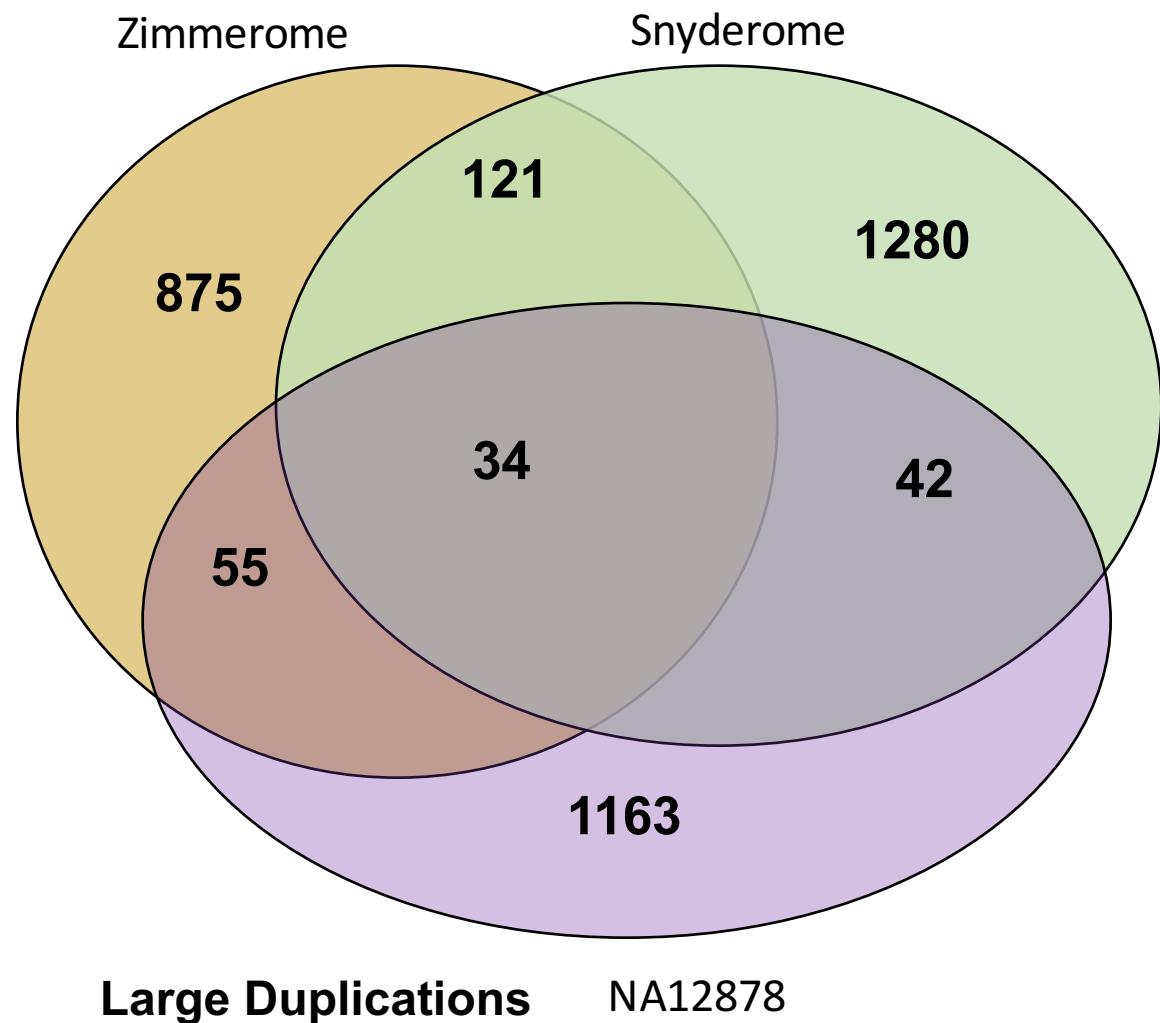
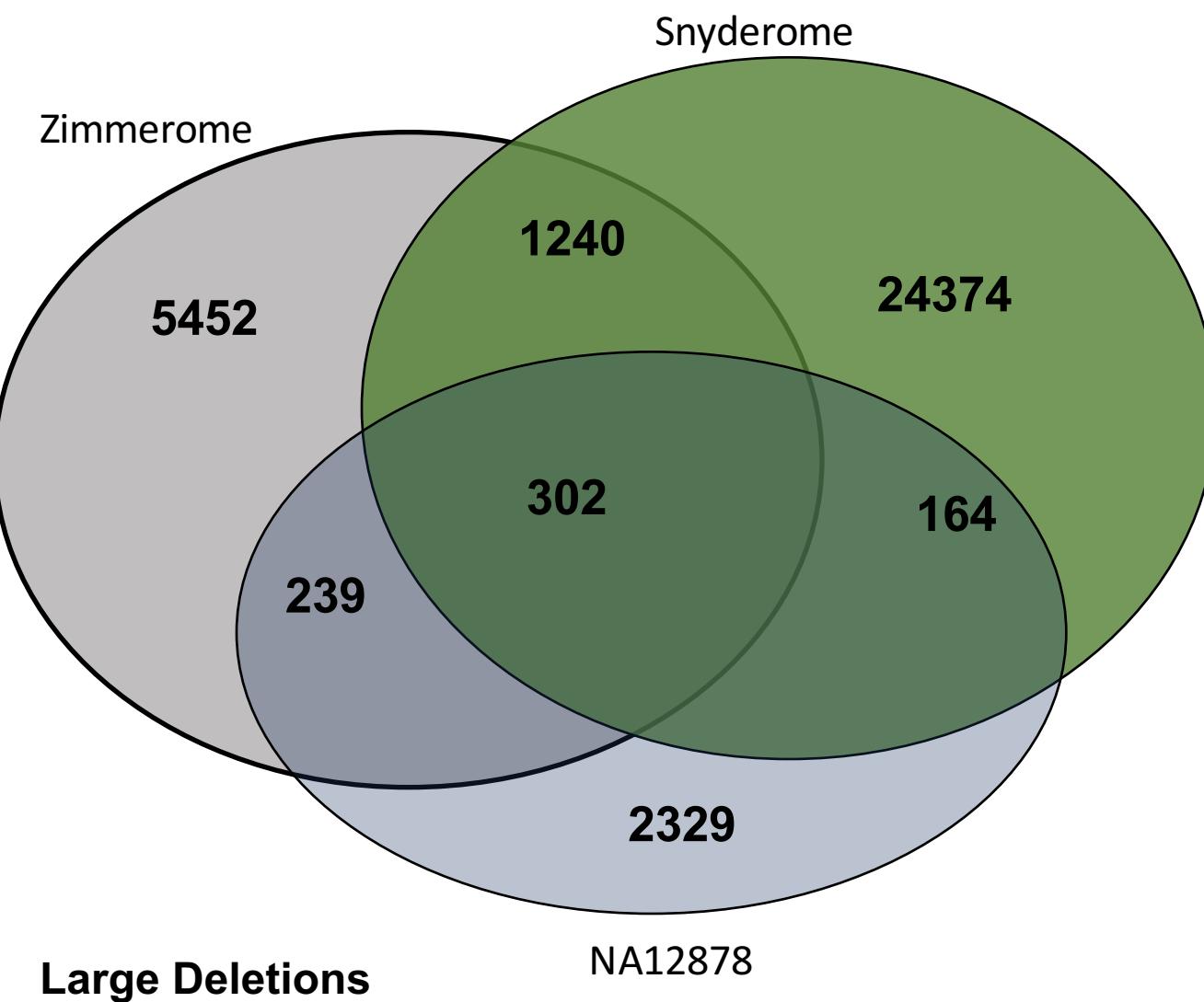
Comparison of calls across three genomes w/o post filtering



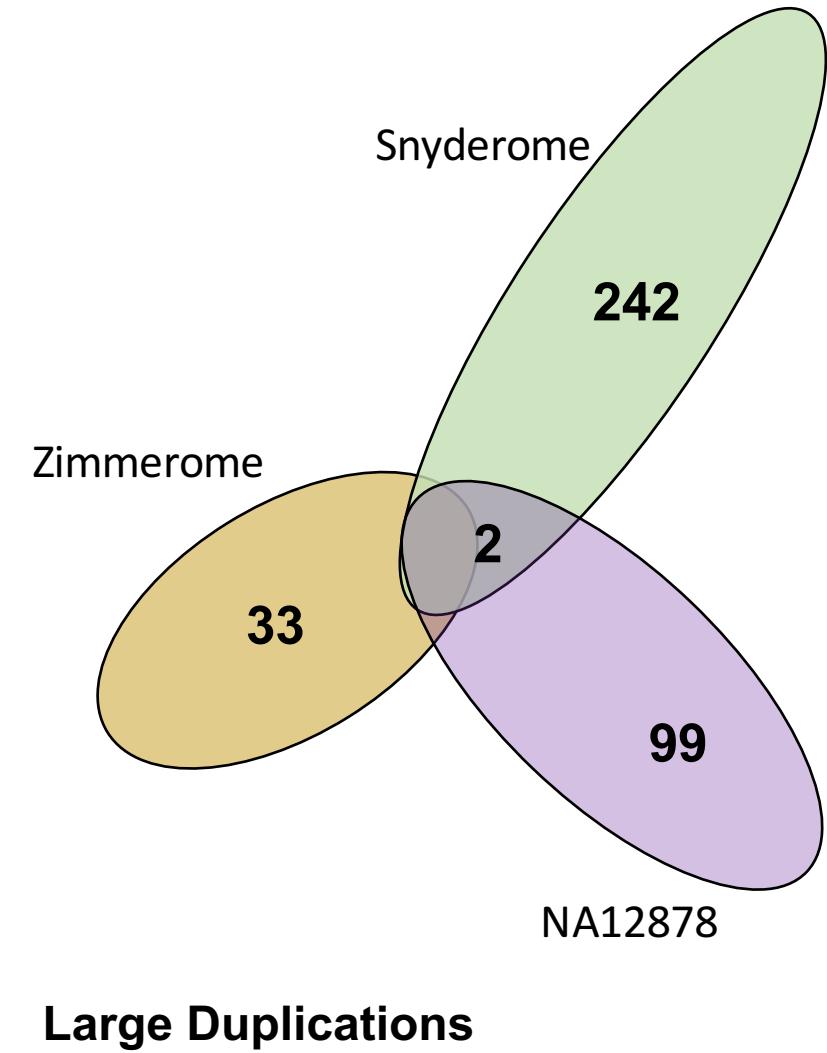
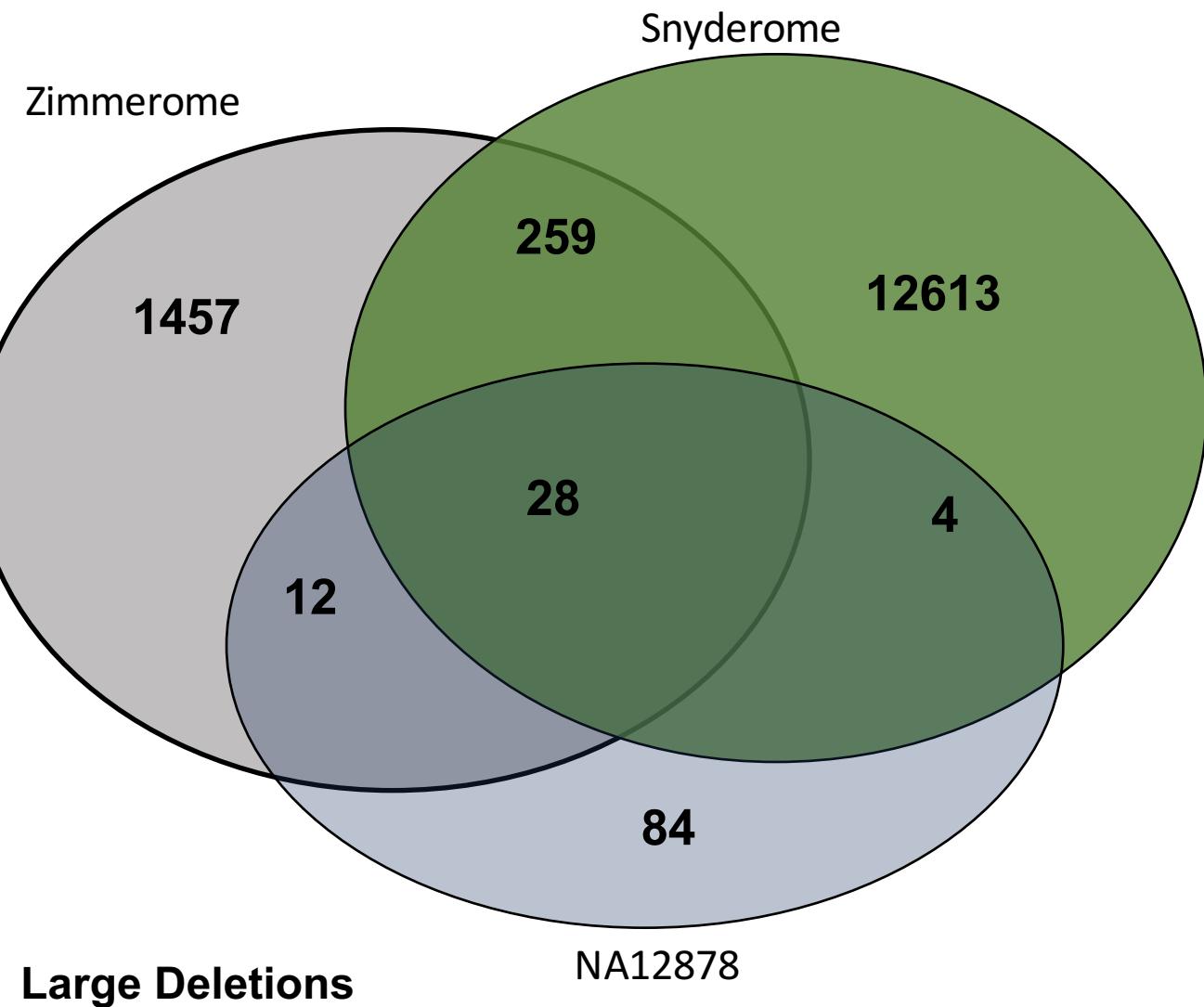
Comparison of calls across three genomes post filtering



Comparison of calls across three genomes w/o post filtering

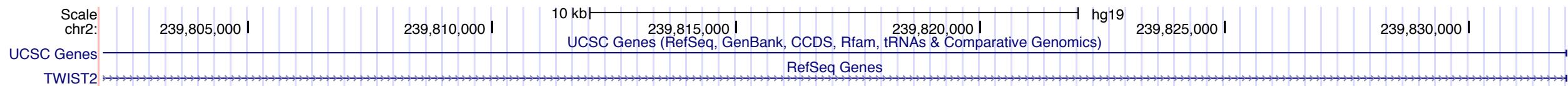


Comparison of calls across three genomes post filtering



Specific examples of Deletions overlapping with various genes

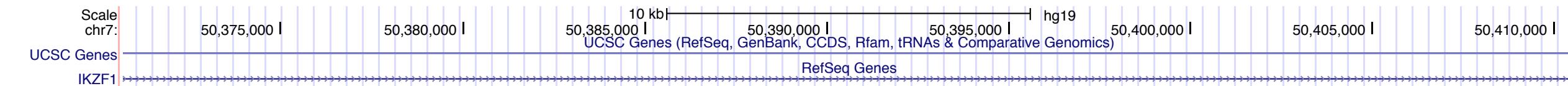
Chr2: 239802001- 239832000 , SVLEN = 30Kb



2q37 deletion syndrome (prominent forehead, highly arched eyebrows, deep-set eyes, a flat nasal bridge, a thin upper lip, and minor ear abnormalities)

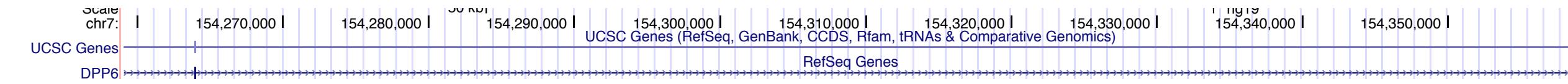
TWIST2 gene : Mutations in this gene cause focal facial dermal dysplasia 3, knock out of this gene in mouse lead to dystonia

Chr7: 50370601- 154370600 , SVLEN = 100Kb



IKZF1 gene : gene associated with B-lymphocyte development and differentiation

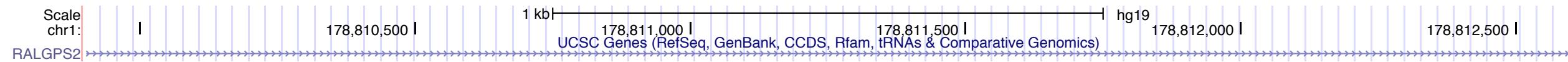
Chr7: 154270601- 50410600 , SVLEN = 40Kb



DPP6 gene : deletion in this gene associated with schizophrenia and attention-deficit hyperactive disorder

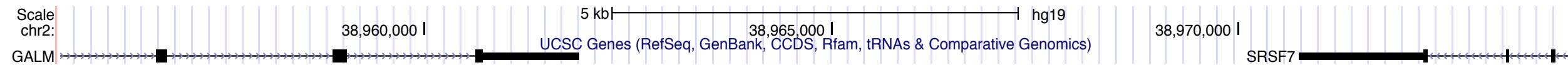
Specific examples of Duplications overlapping with various genes

Chr1: 178809901- 178812600 , SVLEN = 2.7kb



RALGPS2 gene is potential candidate gene for susceptibility to Alzheimer disease

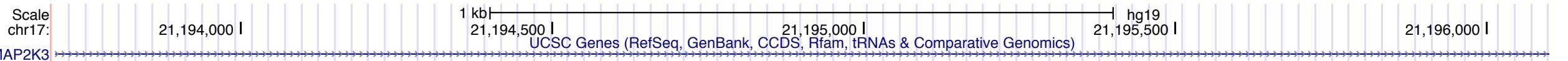
Chr2: 38955501- 38974100 , SVLEN = 18.6Kb



GALM : Creativity in music was found to co-segregate with a duplication covering glucose mutarotase gene

SRSF7 : associated with Allergic Bronchopulmonary Aspergillosis

Chr17: 21193701-21196100 , SVLEN = 2.4Kb



Duplication in chromosome 17p11.2 associated with Potocki-Lupski syndrome