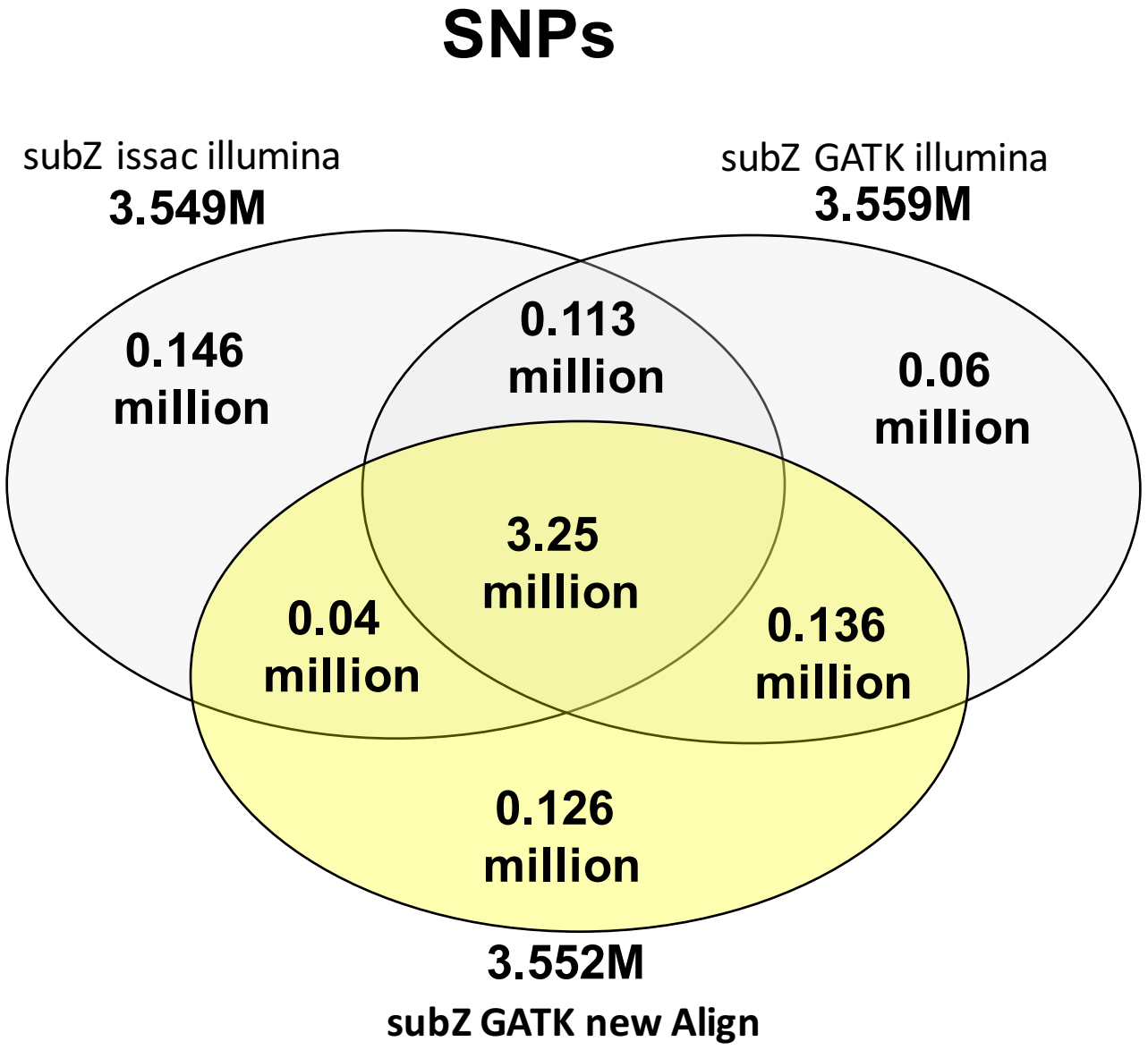
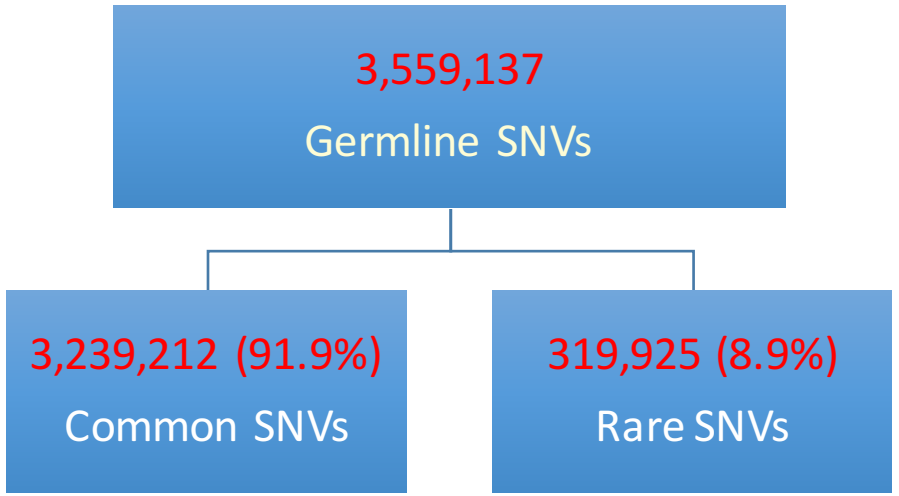
Personal Genome Analysis

Variant calling and Examples

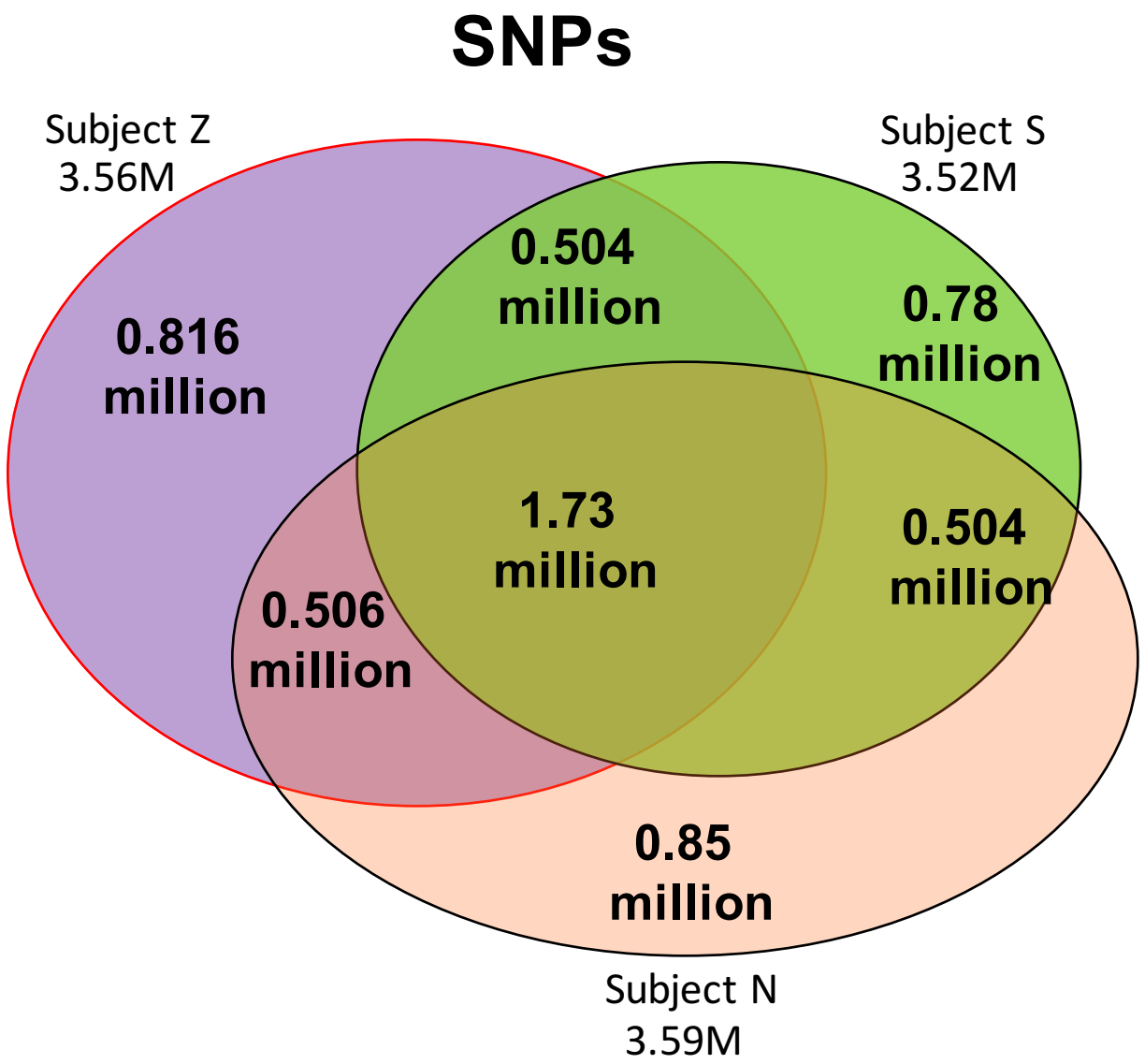
Comparison of variant calls for subject Z

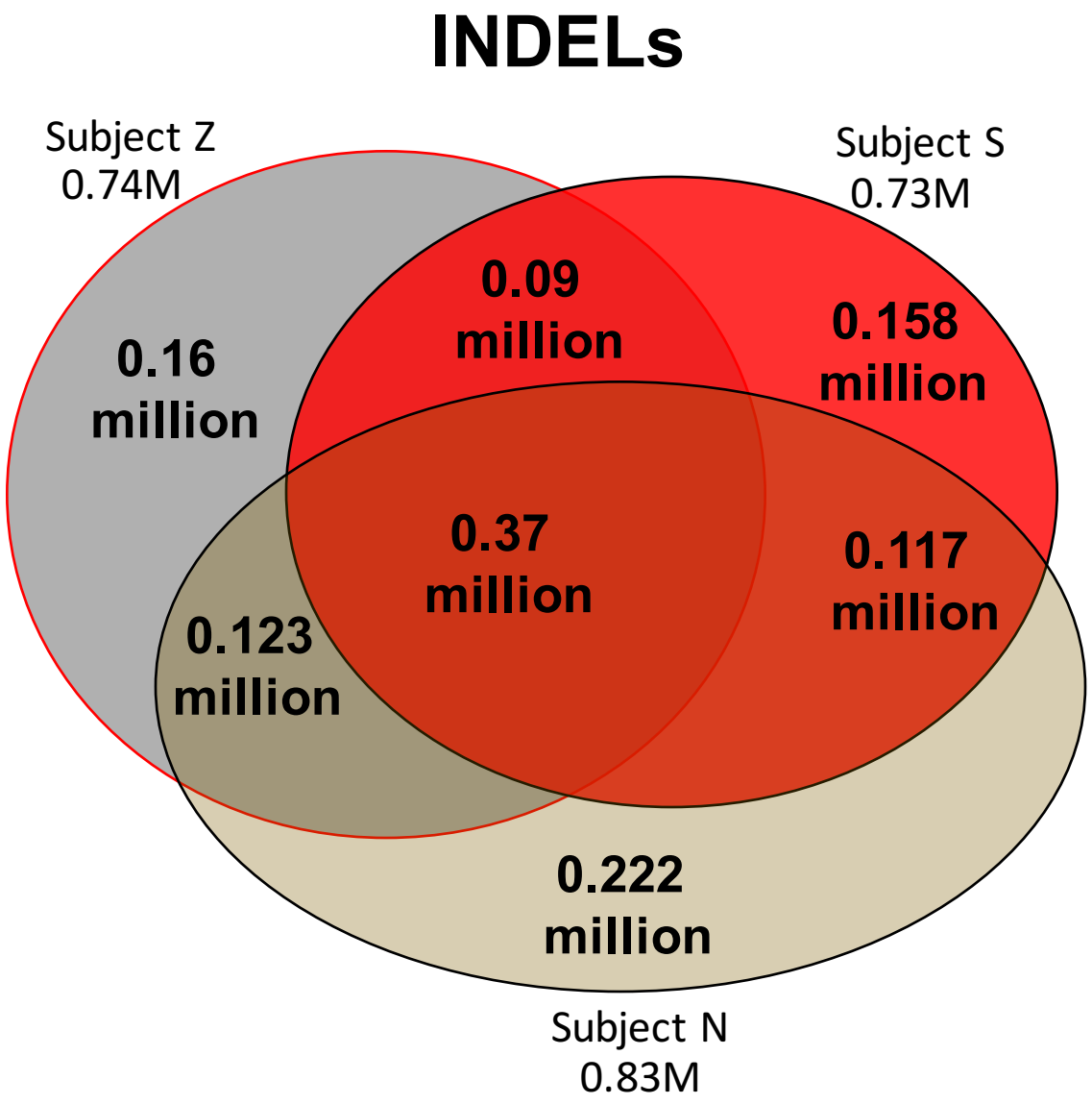


- Original approach (146K specific events):
  - Aligner: CASAVA; Variant Caller: Isaac
- Hybrid approach (60K specific events):
  - Aligner: CASAVA; Variant Caller: GATK
- Gold standard approach (126K specific events):
  - Aligner: BWA; Variant Caller: GATK

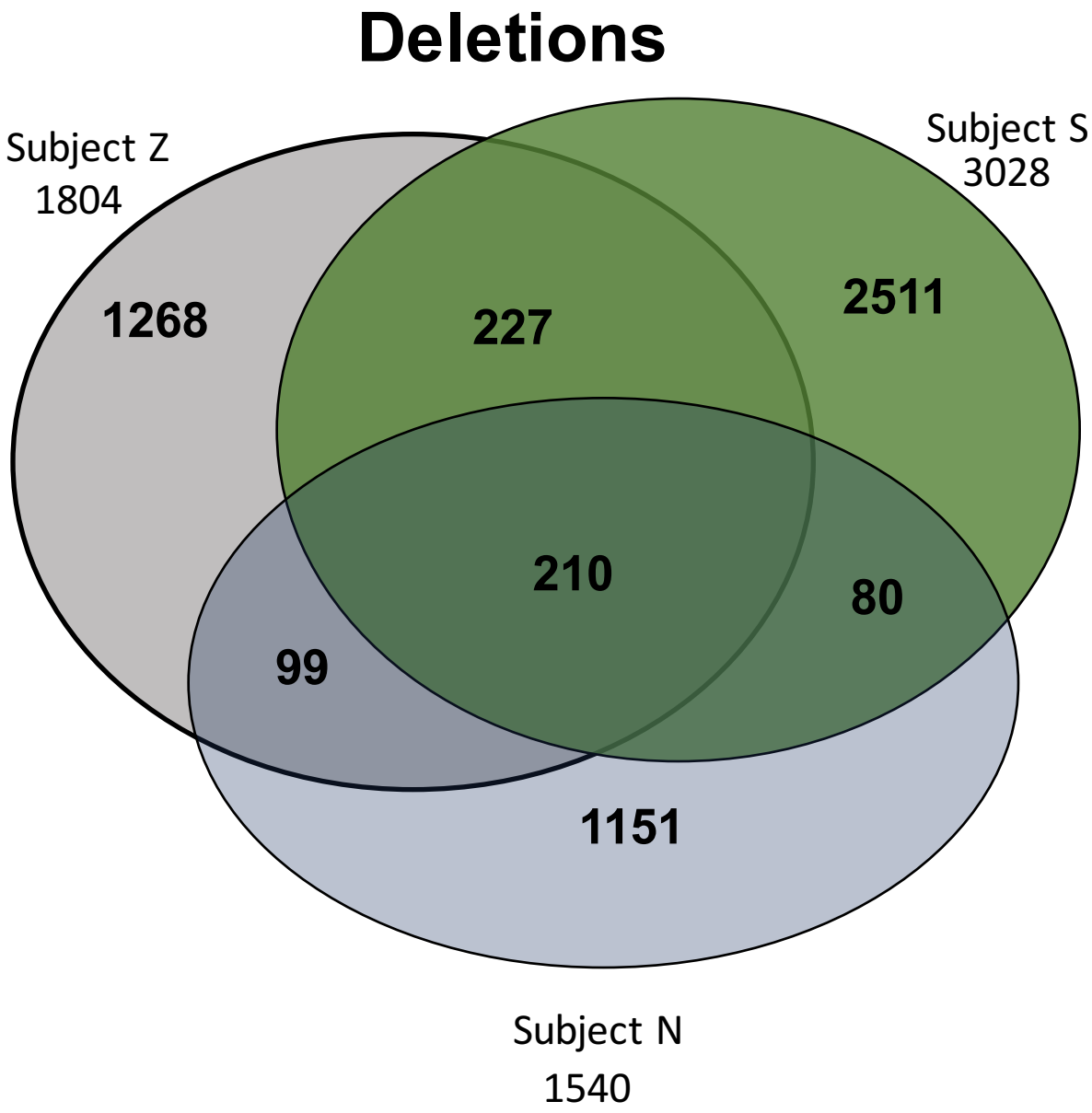


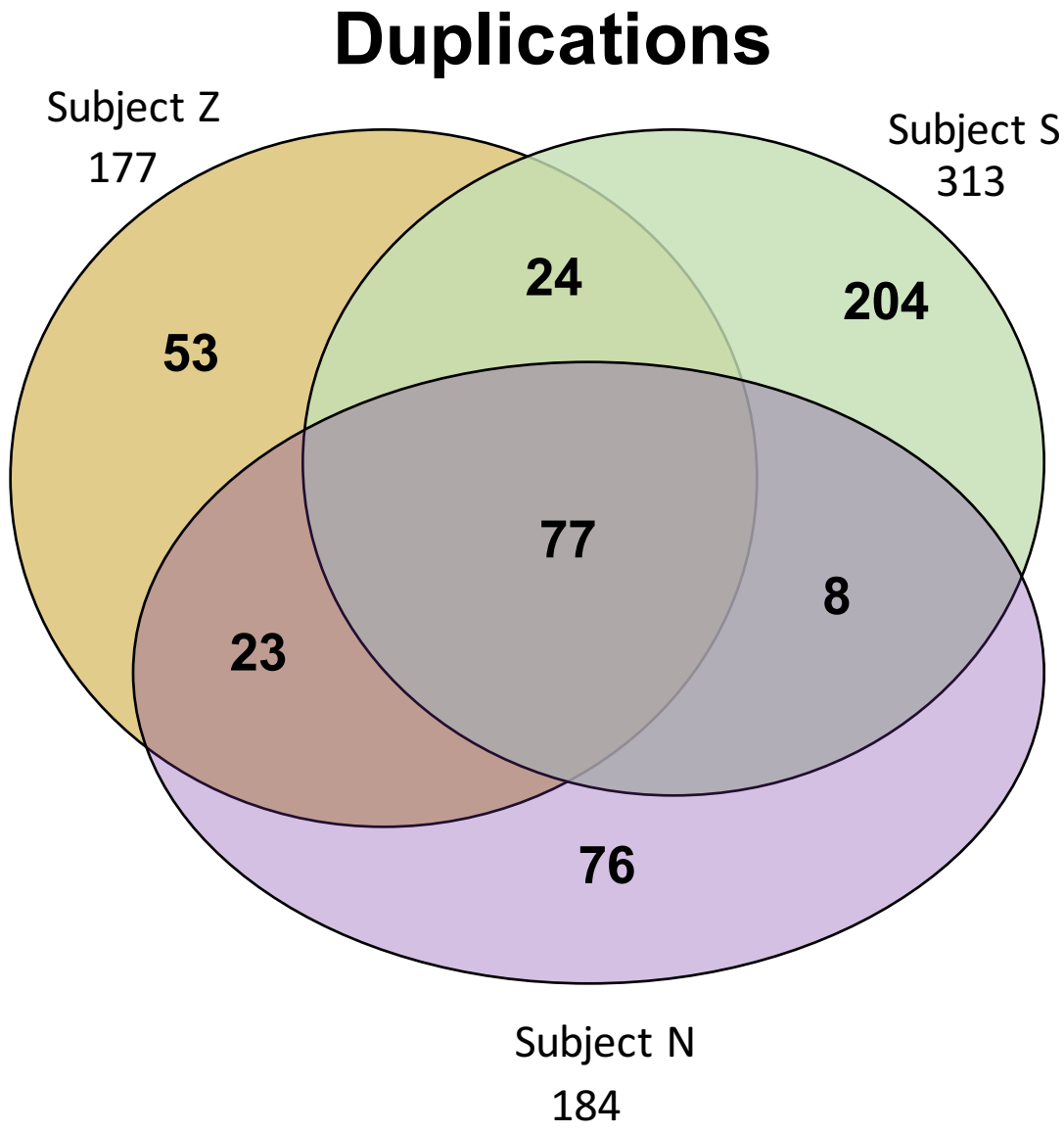
Comparison of SNVs & INDELs across three genomes





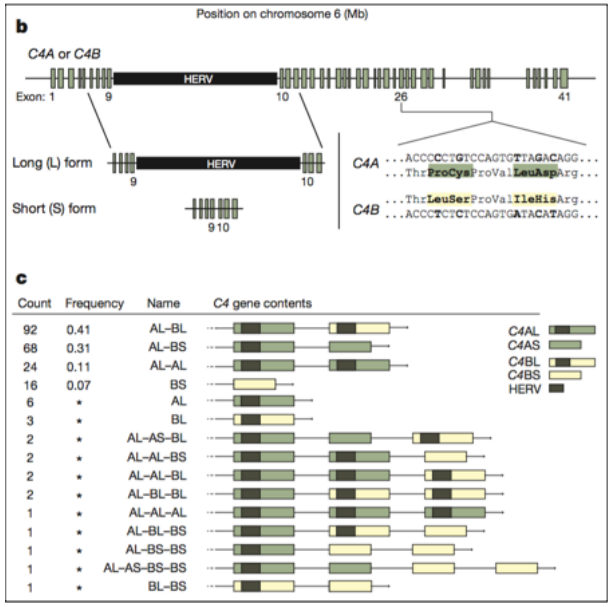
Consensus structural variations across three genomes

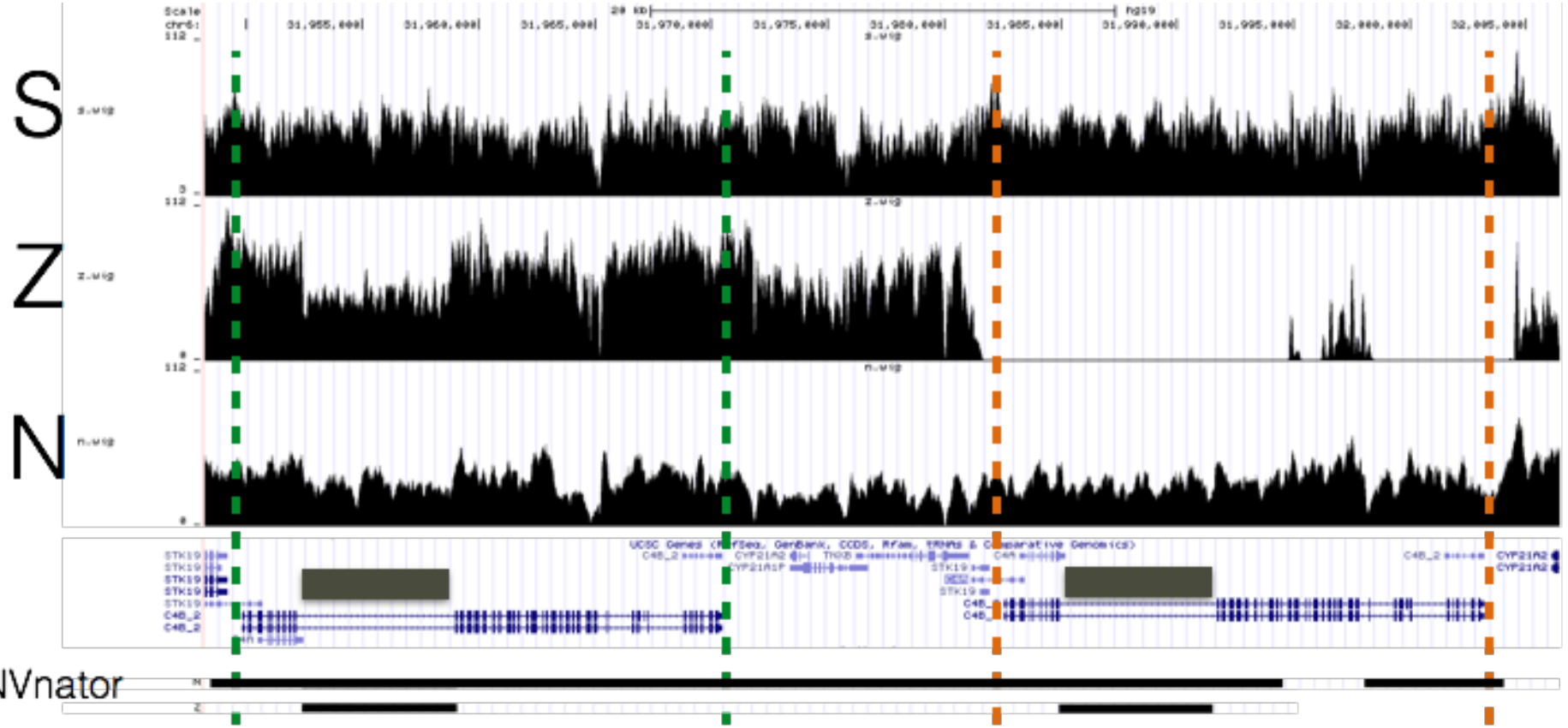


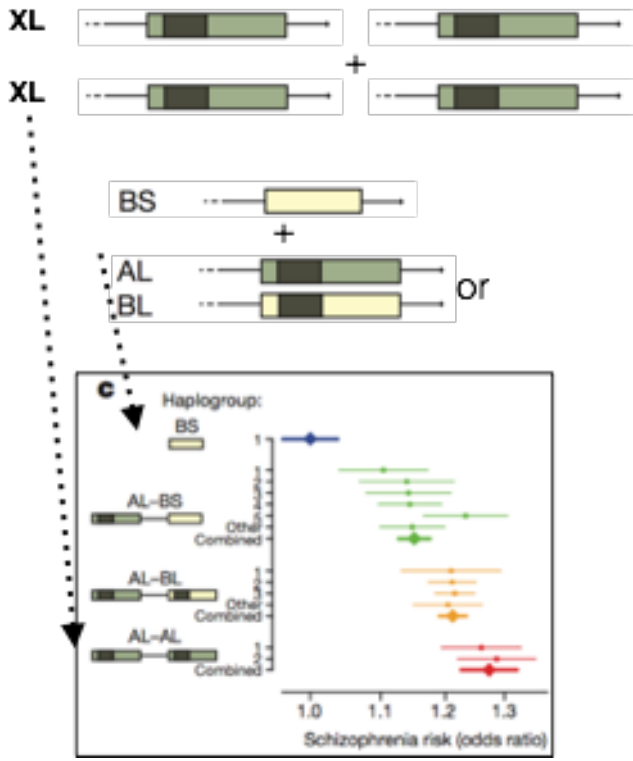


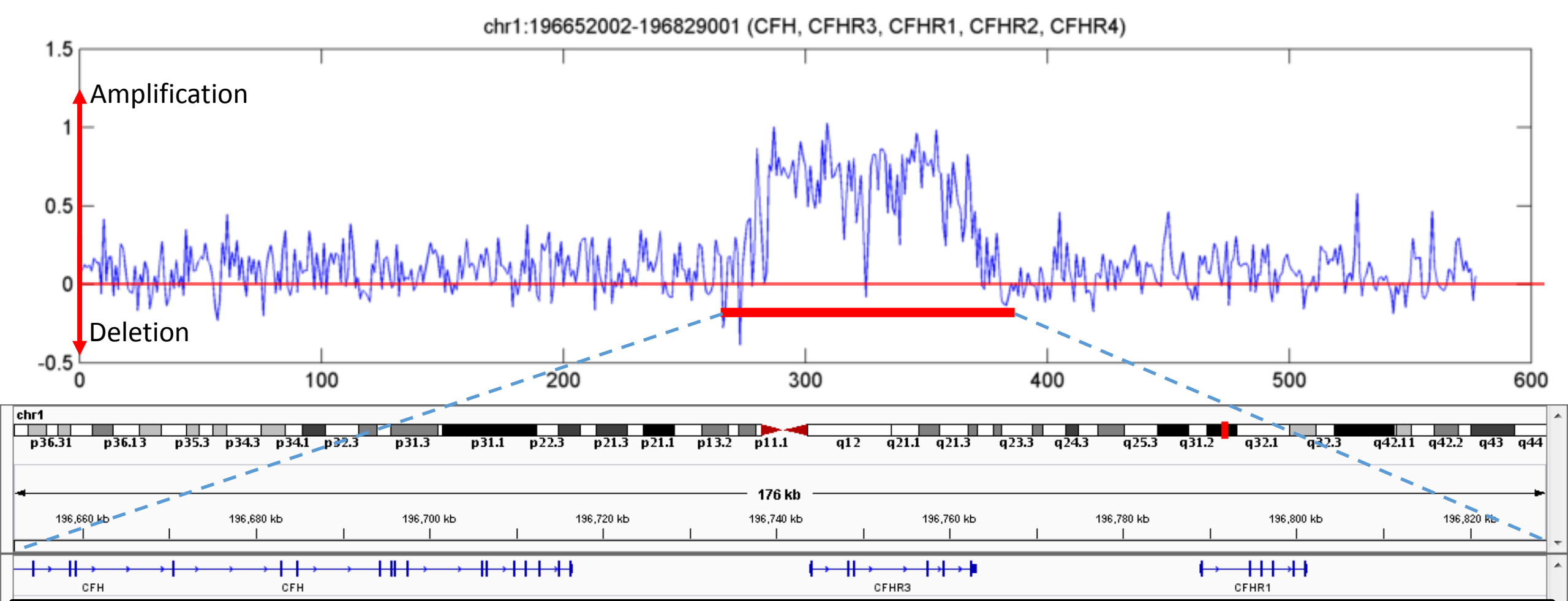
Schizophrenia risk from complex variation of complement component 4

Aswin Sekar[1,2,3], Allison R. Bialas[4,5], Heather de Rivera[1,2], Avery Davis[1,2], Timothy R. Hammond[4], Nolan Kamitaki[1,2], Katherine Tooley[1,2], Jessy Presumey[5], Matthew Baum[1,2,3,4], Vanessa Van Doren[1], Giulio Genovese[1,2], Samuel A. Rose[2], Robert E. Handsaker[1,2], Schizophrenia Working Group of the Psychiatric Genomics Consortium*, Mark J. Daly[2,6], Michael C. Carroll[5], Beth Stevens[2,4] & Steven A. McCarroll[1,2]



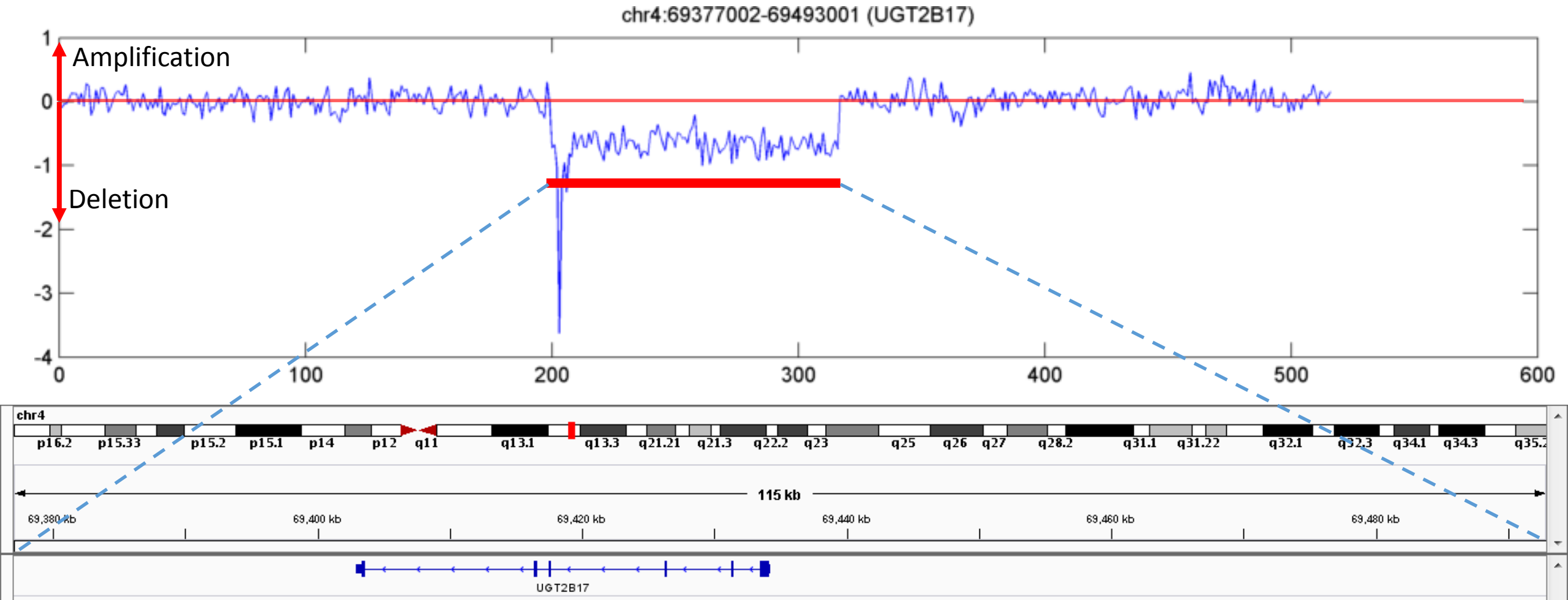






A common *CFH* haplotype, with deletion of *CFHR1* and *CFHR3*, is associated with lower risk of age-related macular degeneration

Anne E Hughes[1], Nick Orr[1], Hossein Esfandiary[1], Martha Diaz-Torres[2], Timothy Goodship[2] & Usha Chakravarthy[3]



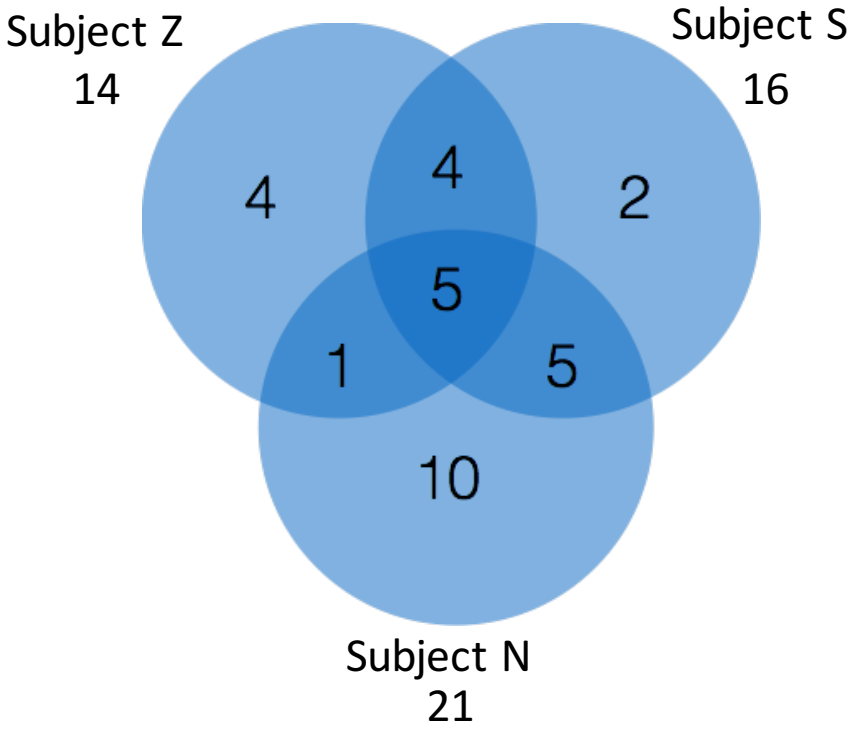
Deletion Polymorphism of UDP-Glucuronosyltransferase 2B17 and Risk of Prostate Cancer in African American and Caucasian Men

can American controls, respectively. When all subjects were considered, a significant association was found between the *UGT2B17* deletion polymorphism and prostate cancer risk

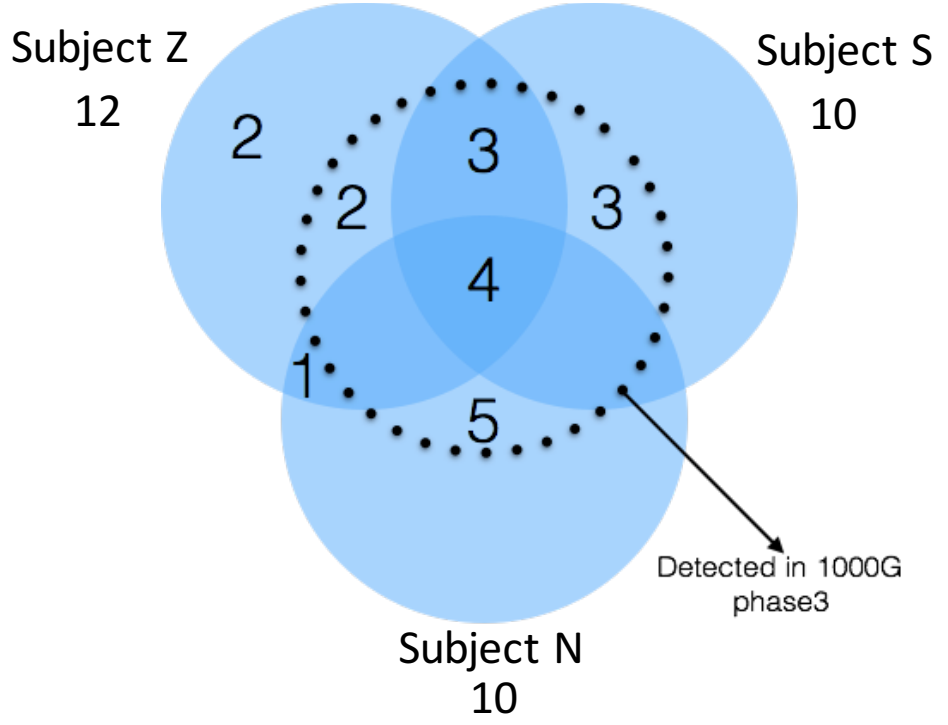
Processed Pseudogene CNV

	Pseudogenes	Processed pseudogenes	Human specific processed pseudogenes
Human	~14,000	7,831	127

Pseudogene absence

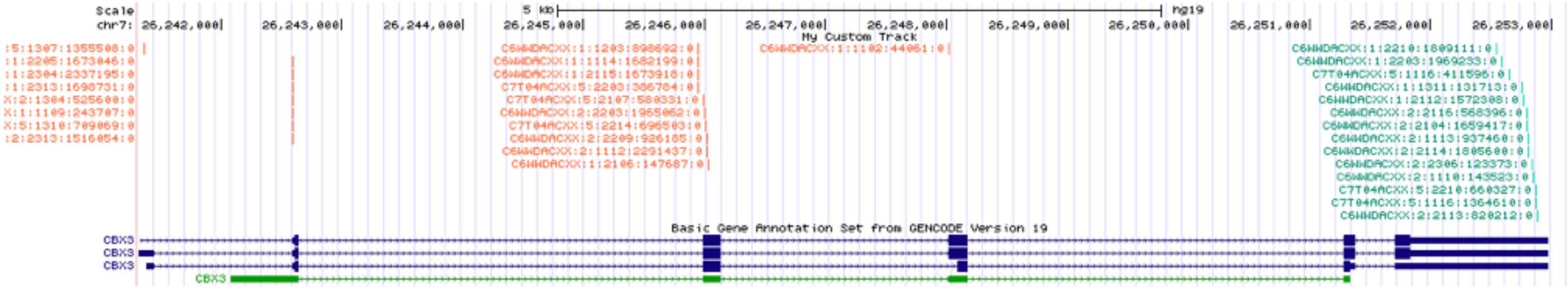


Pseudogene insertion



Pseudogene CNV – Example I

CBX3 Parental gene



CBX3 Insertion Point

ALU variation

	# of Alu in the genome	AluY
Human	1,238,995	146,308