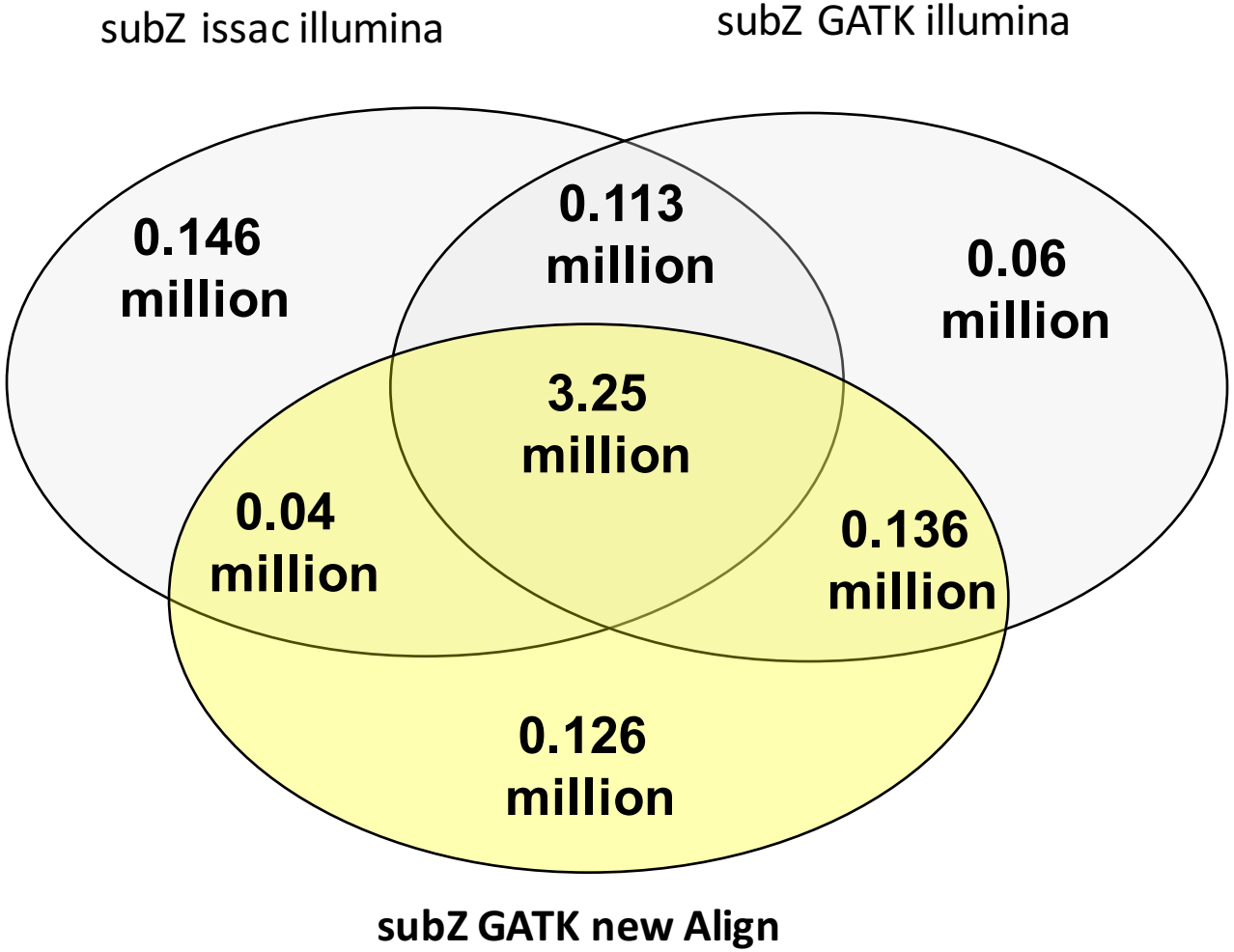
Personal Genome Analysis

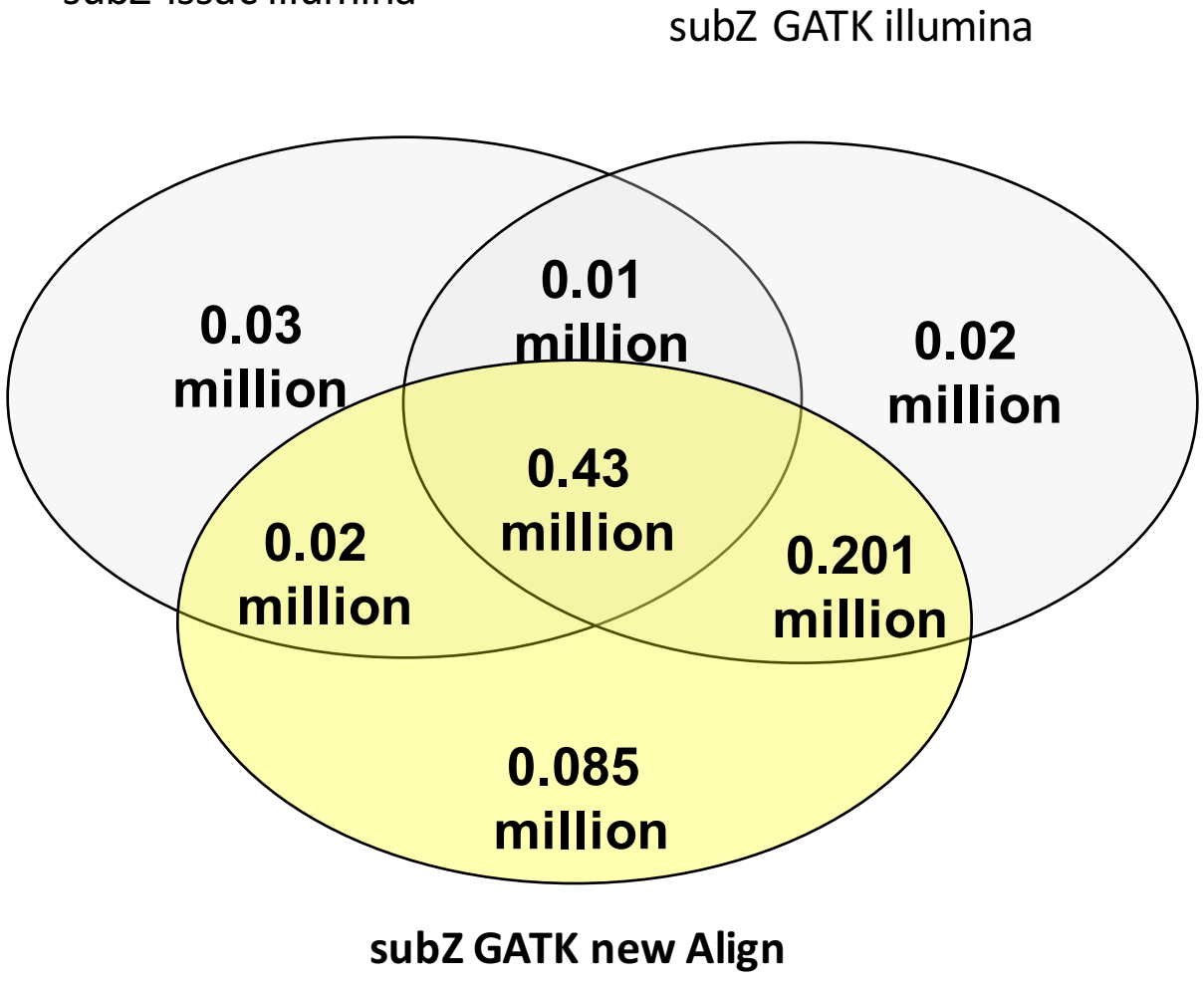
Variant calling and Examples

Comparison of variant calls for subject Z

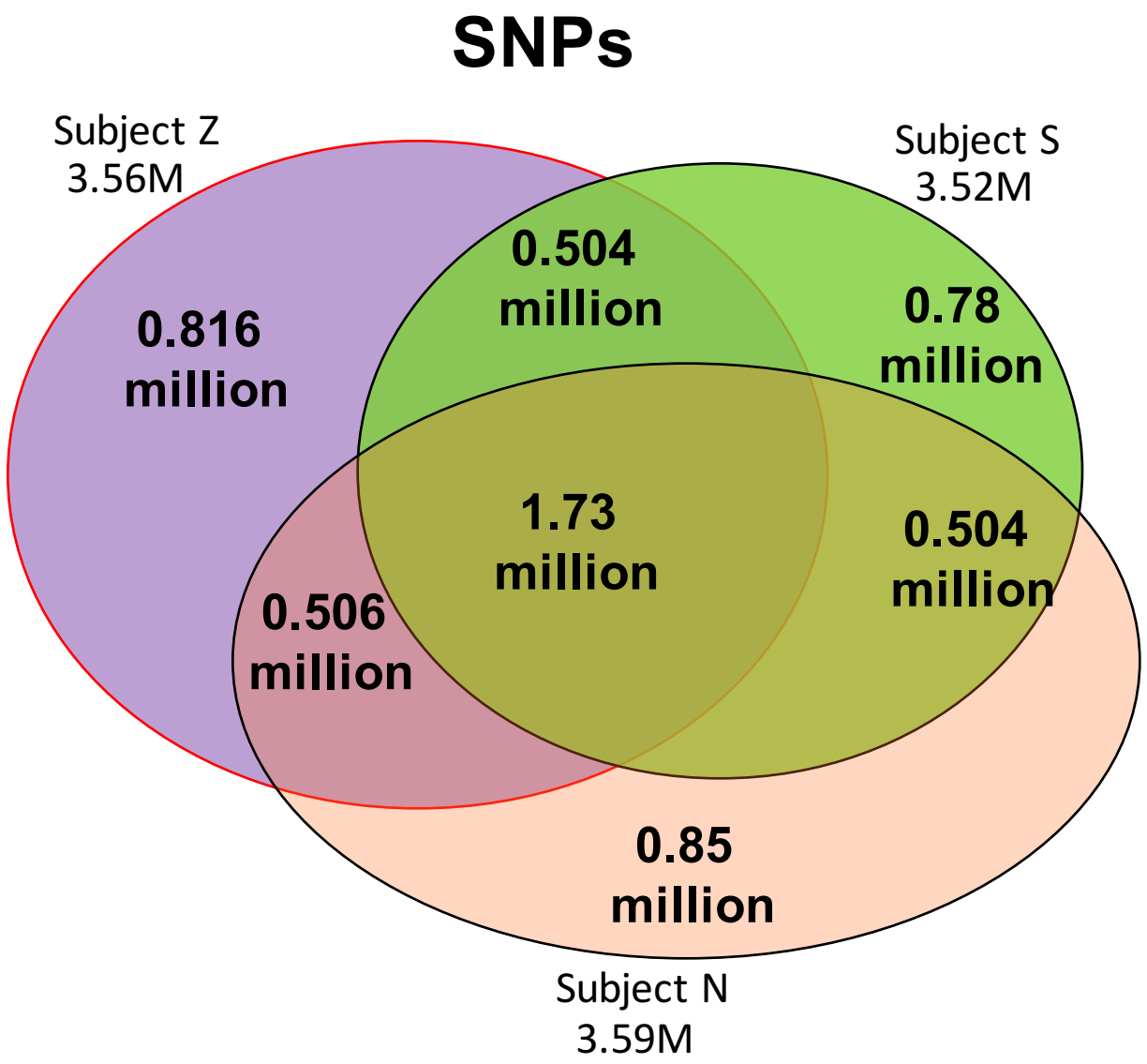
SNPs

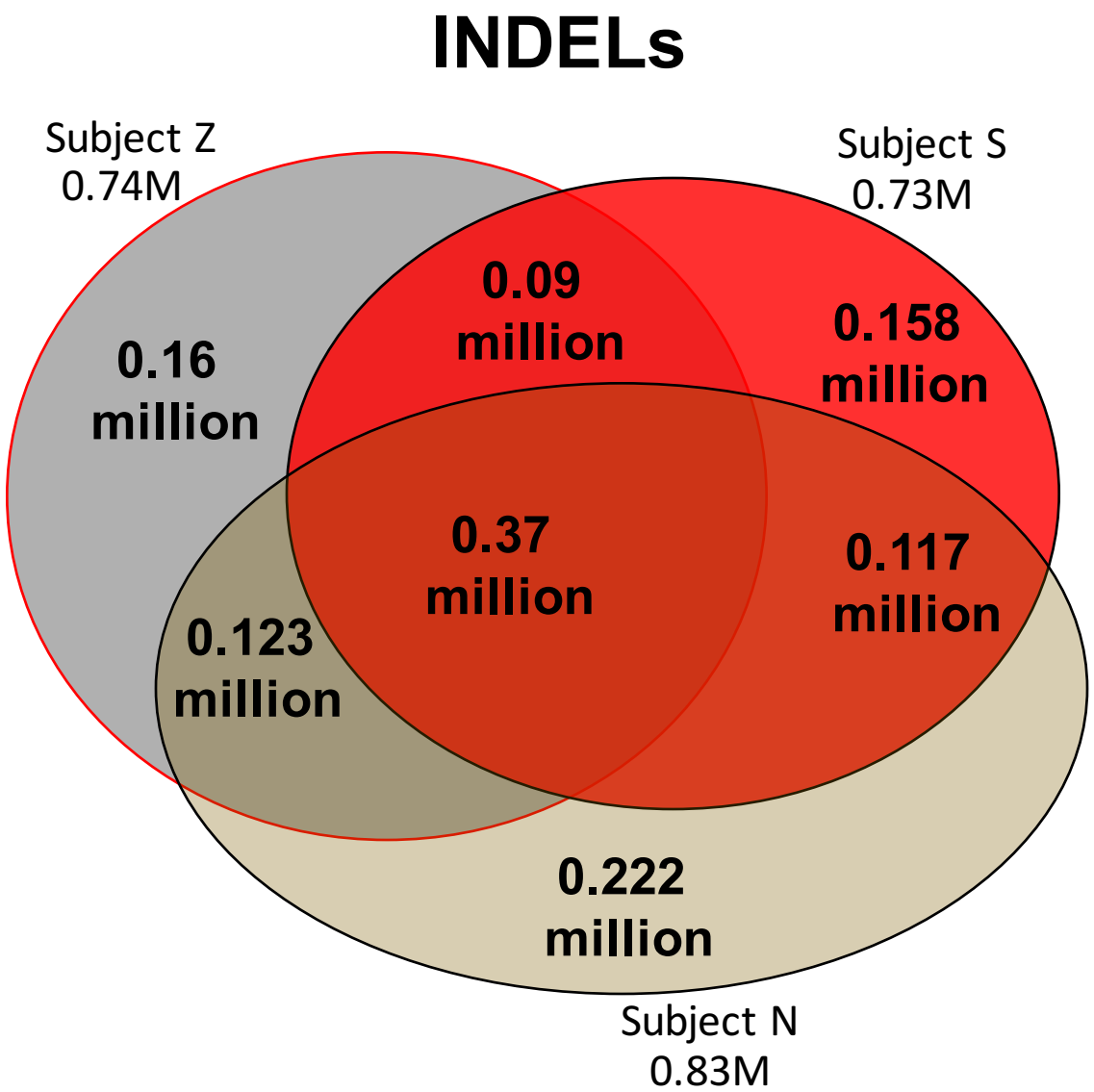


INDELs

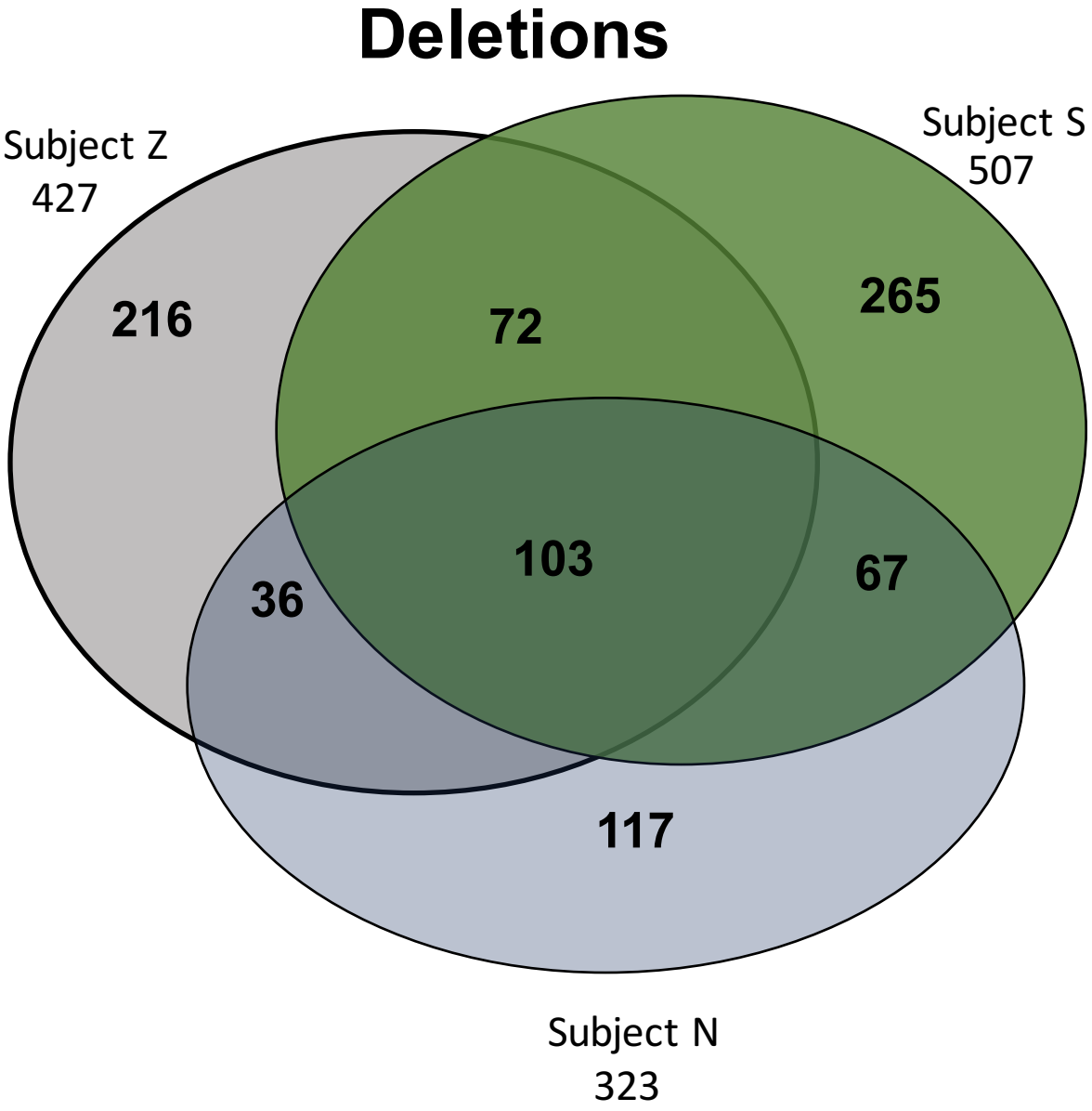


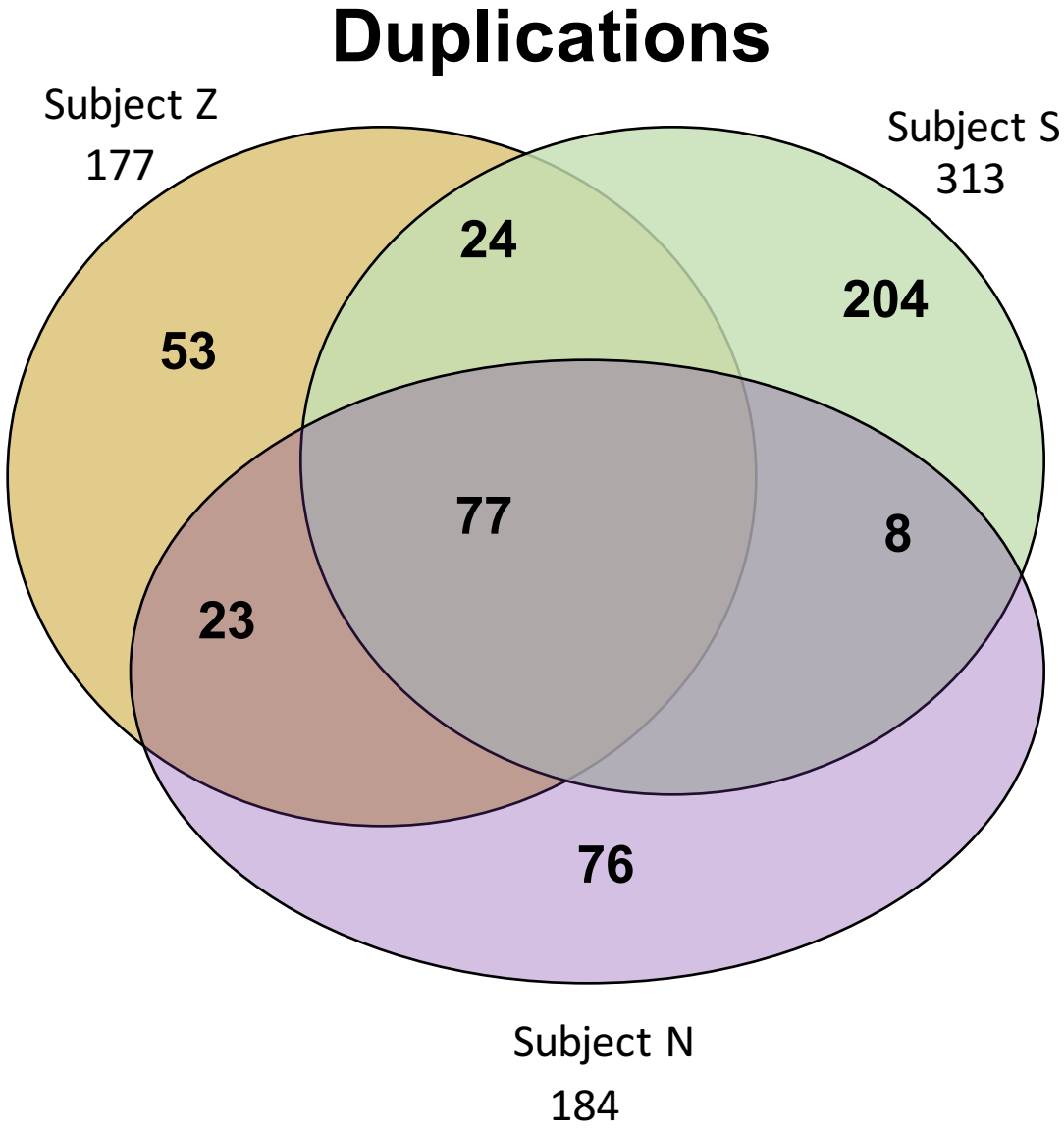
Comparison of SNVs & INDELs across three genomes





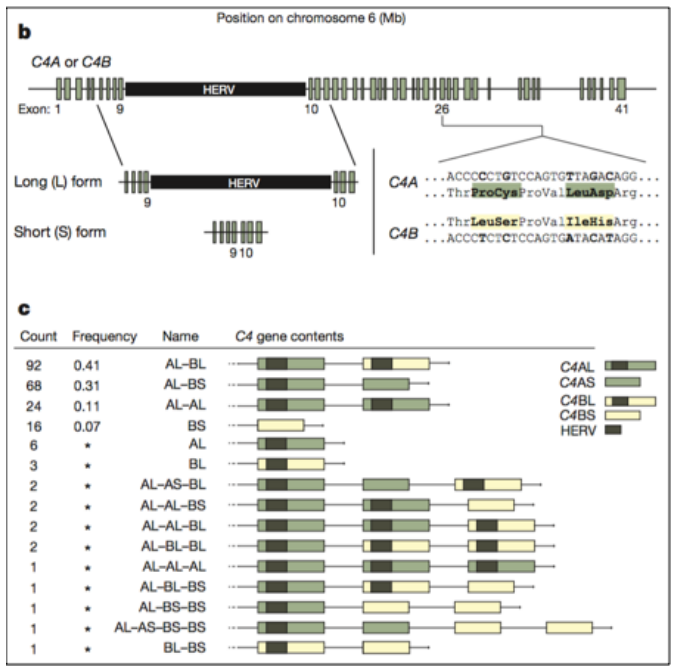
Consensus structural variations across three genomes

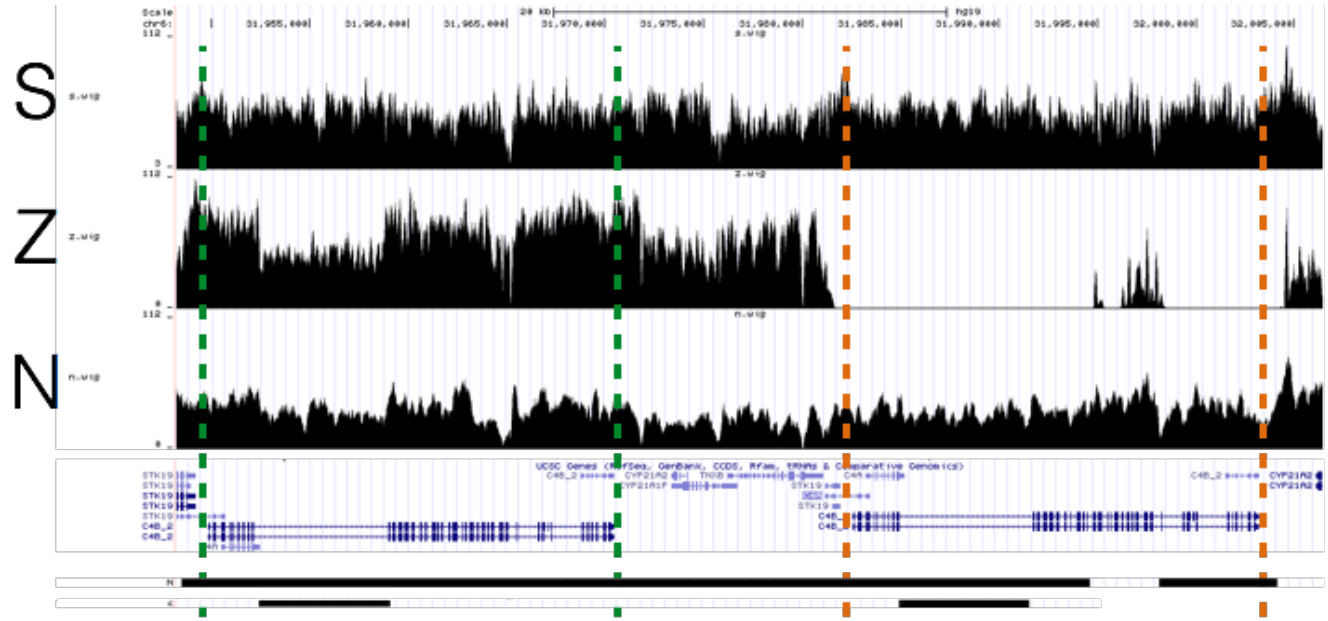


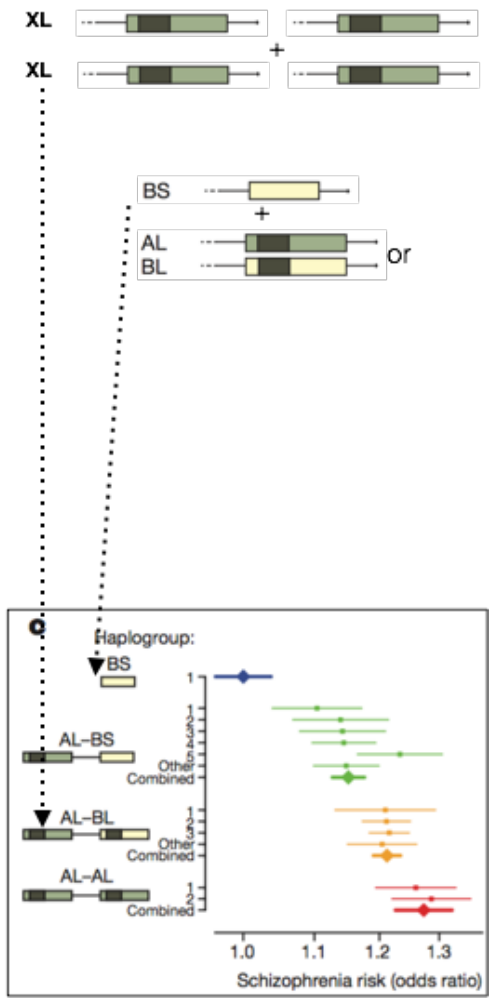


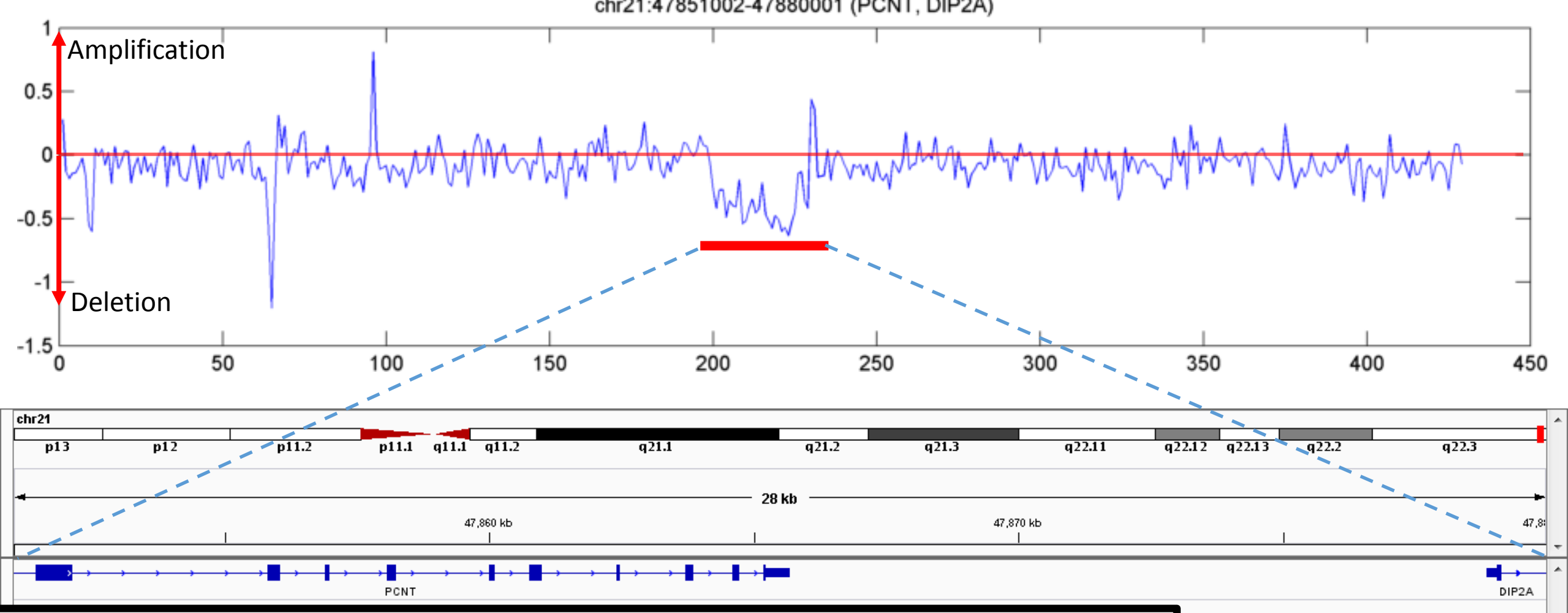
Schizophrenia risk from complex variation of complement component 4

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]





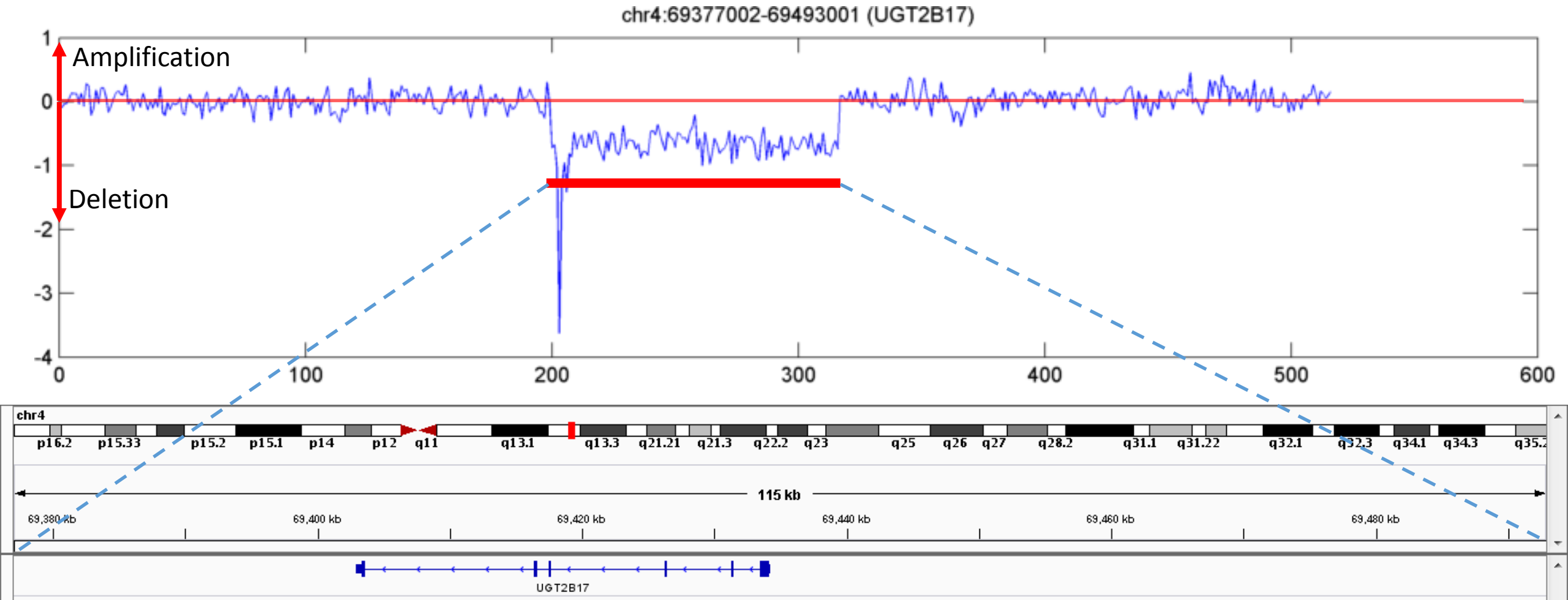




Identification of Novel Dyslexia Candidate Genes Through the Analysis of a Chromosomal Deletion

G. Poelmans,[1,2]* J.J.M. Engelen,[1,3] J. Van Lent-Albrechts,[1]
B. Franke,[4,5] J.K. Buitelaar,[5] M. Wuisman-Frerker,[6] W. Erer
and C. Schrander-Stumpel[1,3]

this article, we report the identification of four new dyslexia candidate genes (*PCNT*, *DIP2A*, *S100B*, and *PRMT2*) on chromosome region 21q22.3 by FISH and SNP microarray analyses of a very small deletion in this region, which cosegregates with



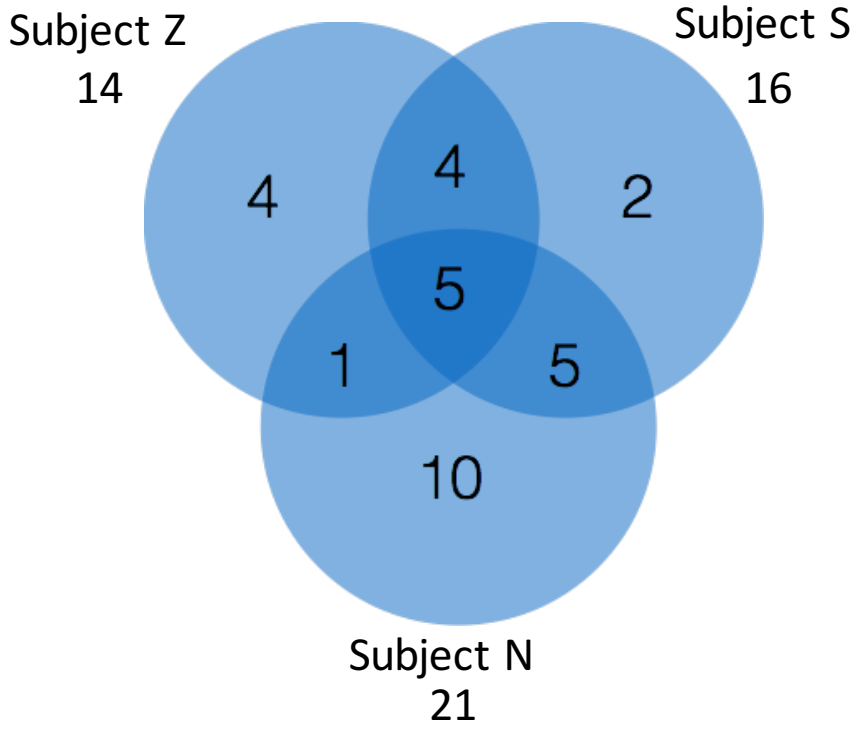
Deletion Polymorphism of UDP-Glucuronosyltransferase 2B17 and Risk of Prostate Cancer in African American and Caucasian Men

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

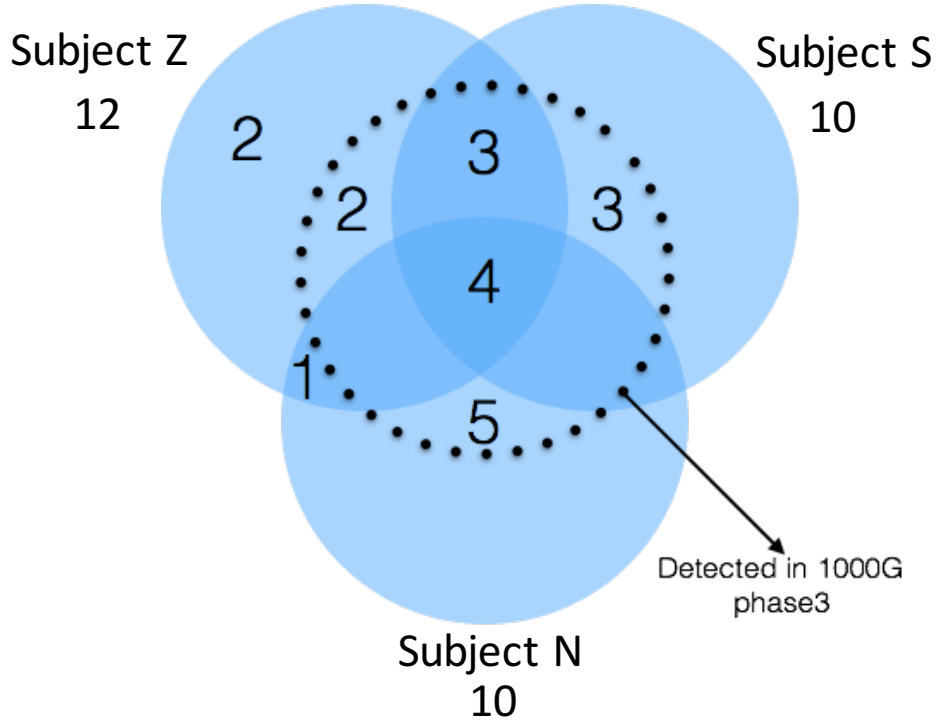
Pseudogene CNV

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

Pseudogene absence

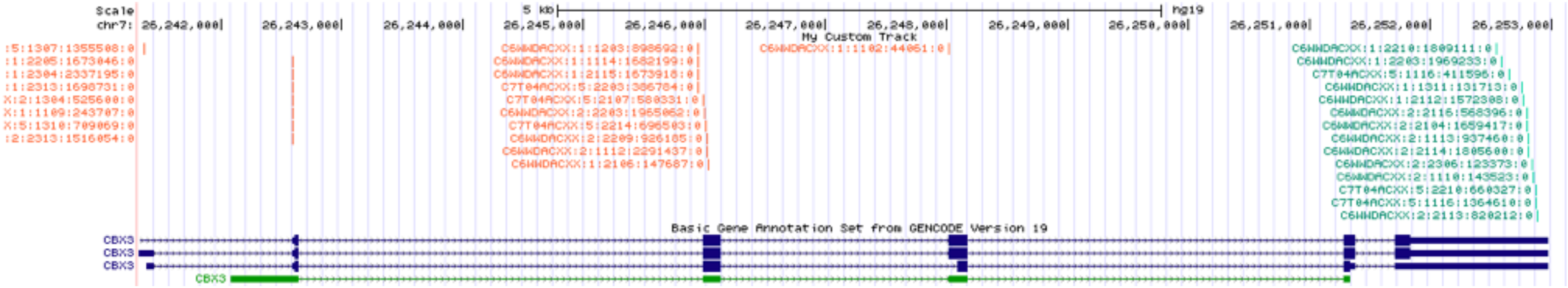


Pseudogene insertion



Pseudogene CNV – Example I

CBX3 Parental gene



CBX3 Insertion Point

ALU variation

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