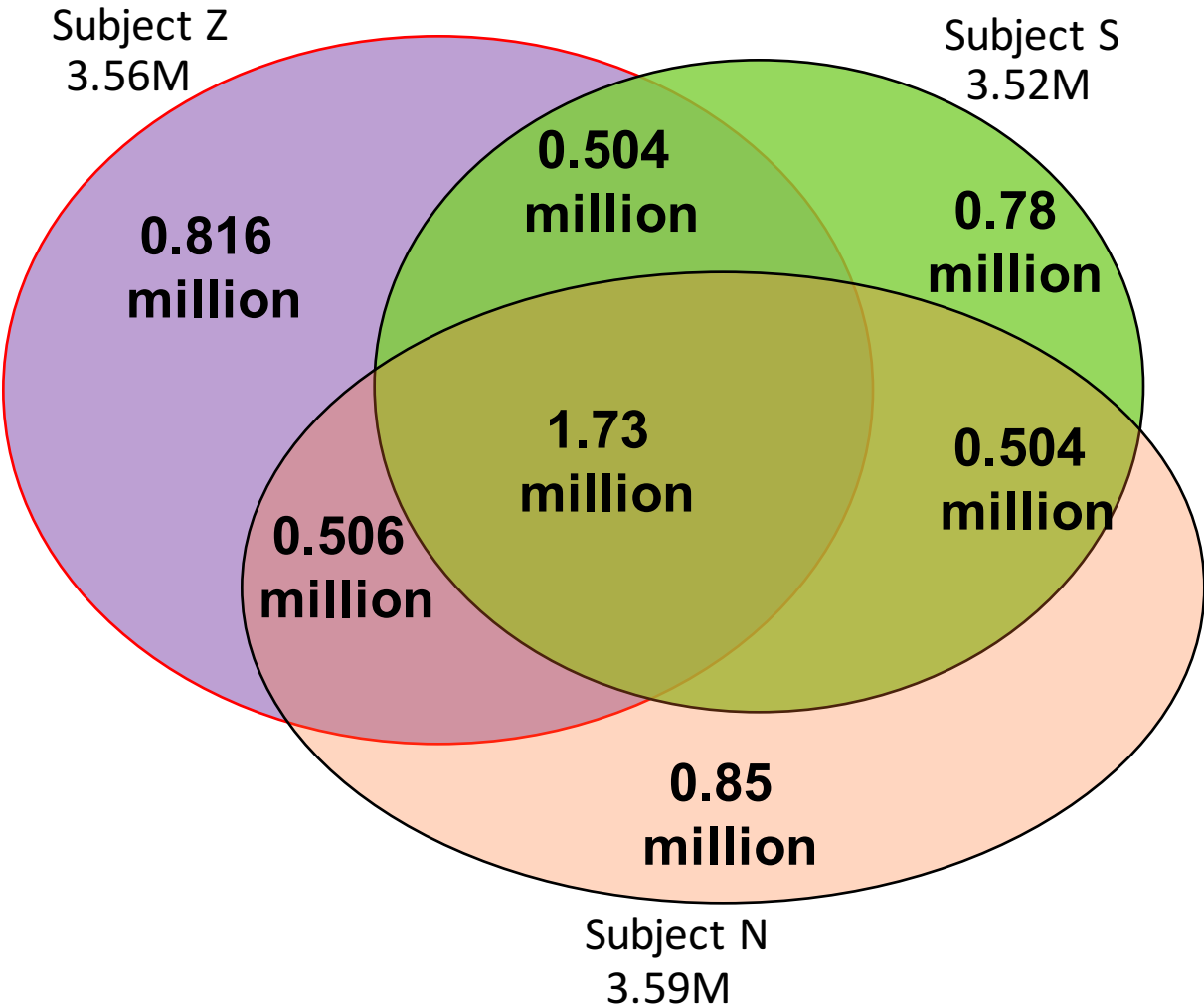


# Personal Genome Analysis

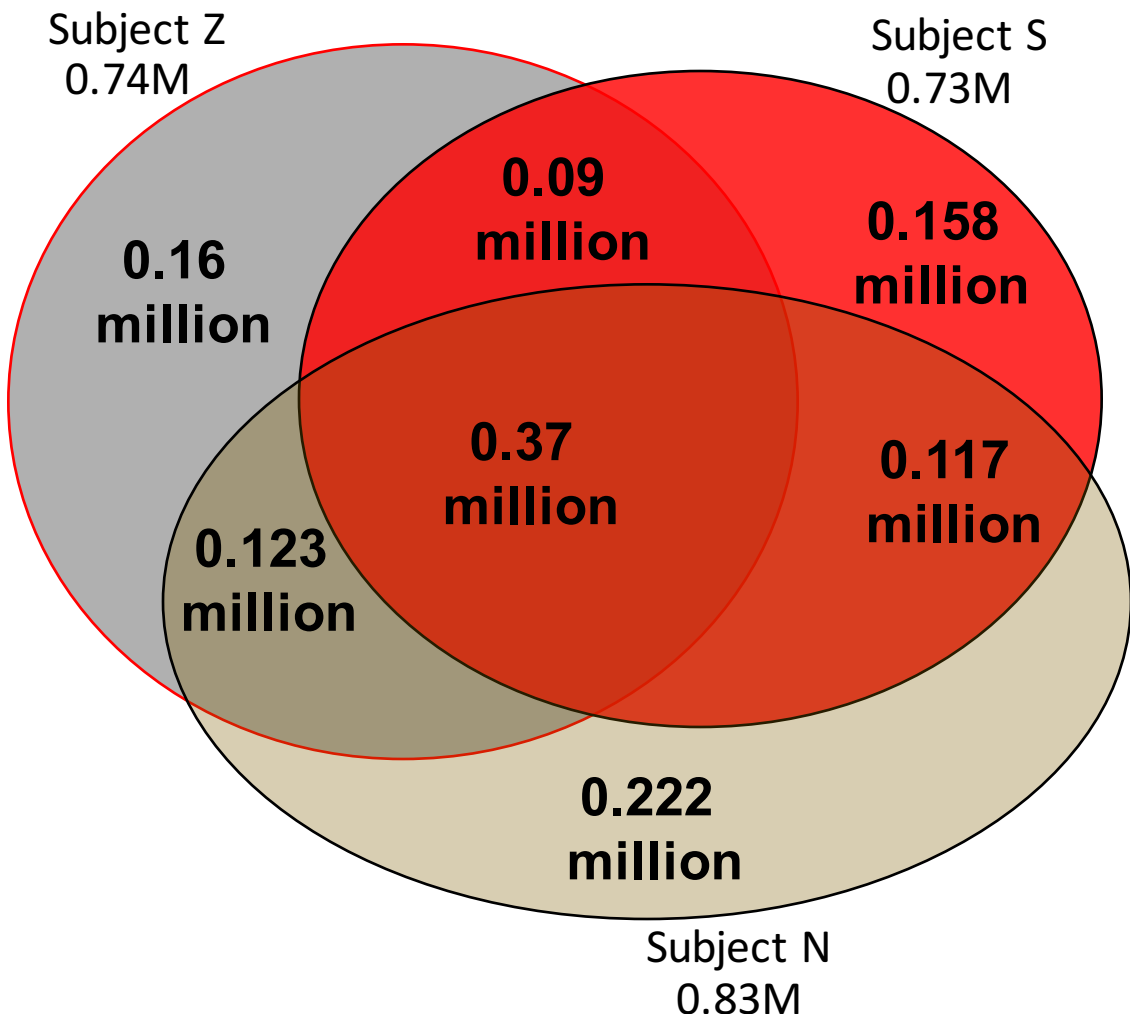
Variant calling and Examples

# Comparison of SNVs & INDELs across three genomes

## SNPs

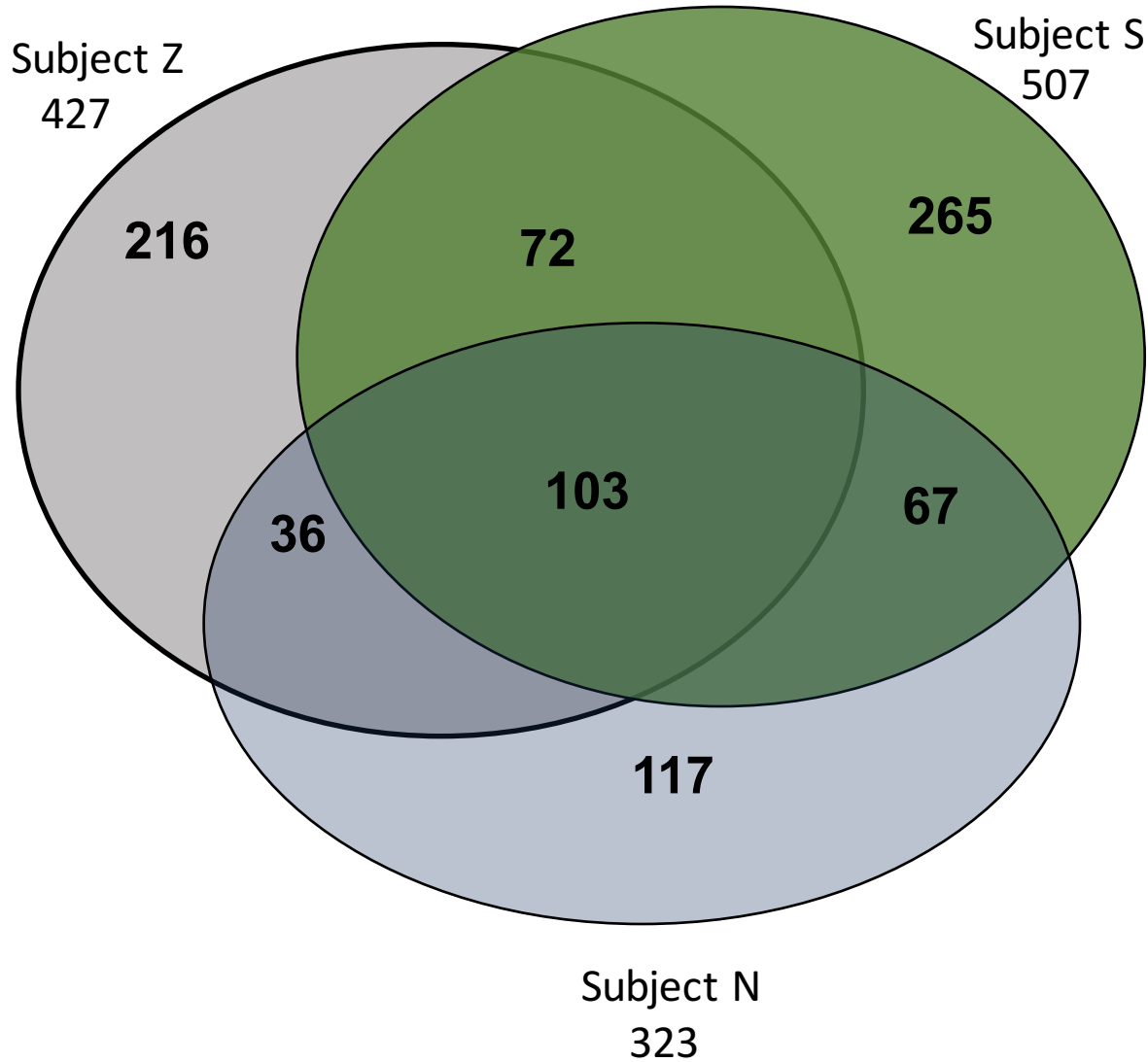


## INDELs

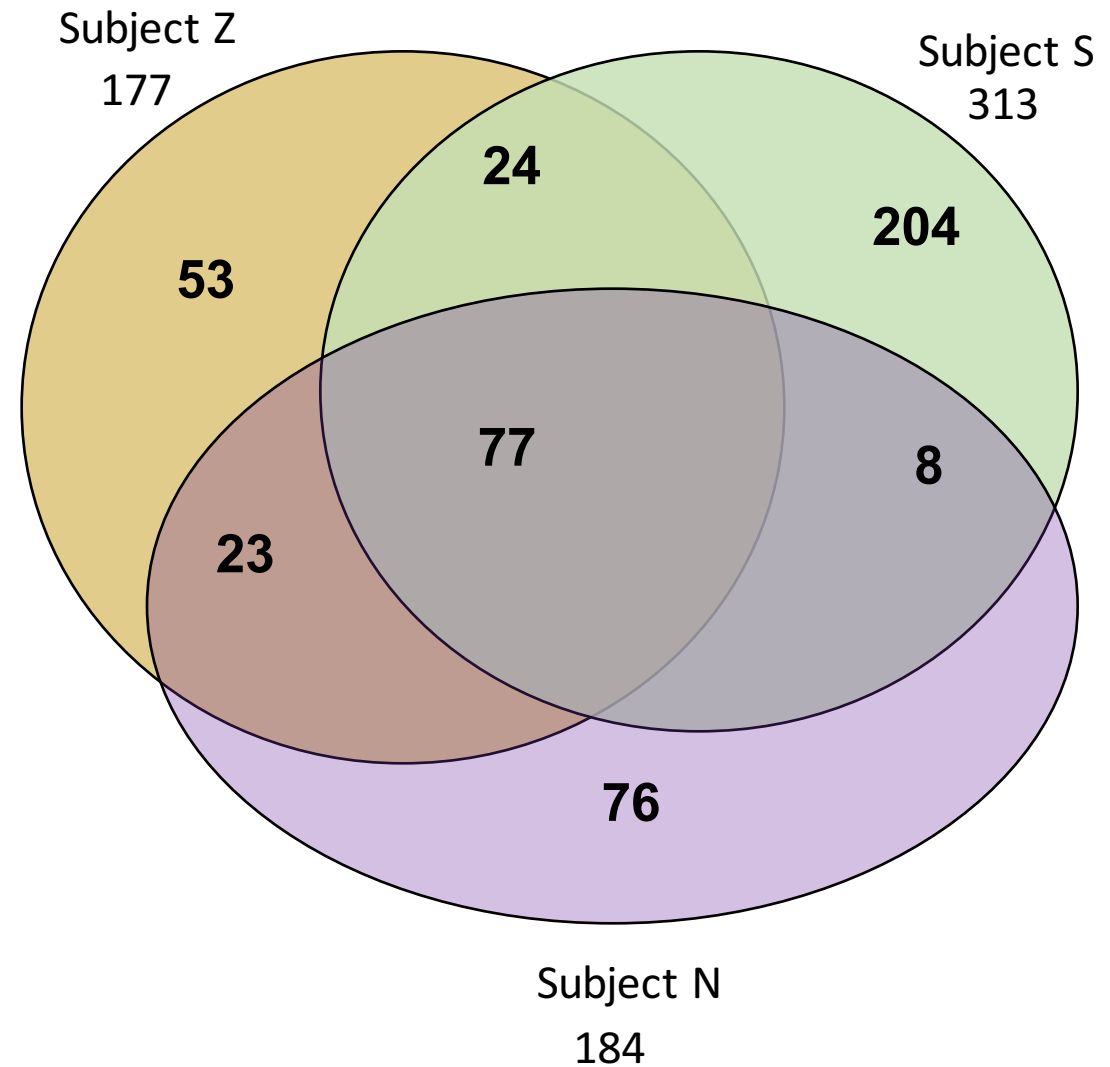


# Consensus structural variations across three genomes

## Deletions

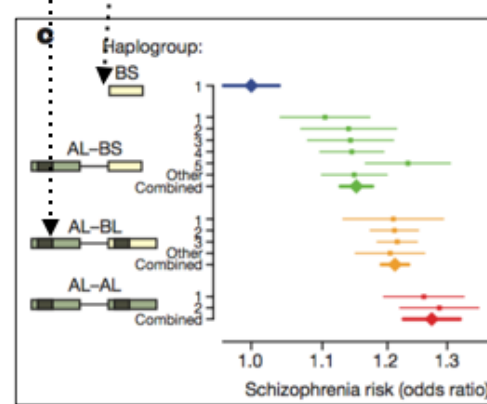
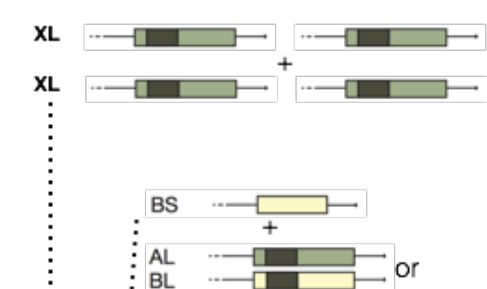
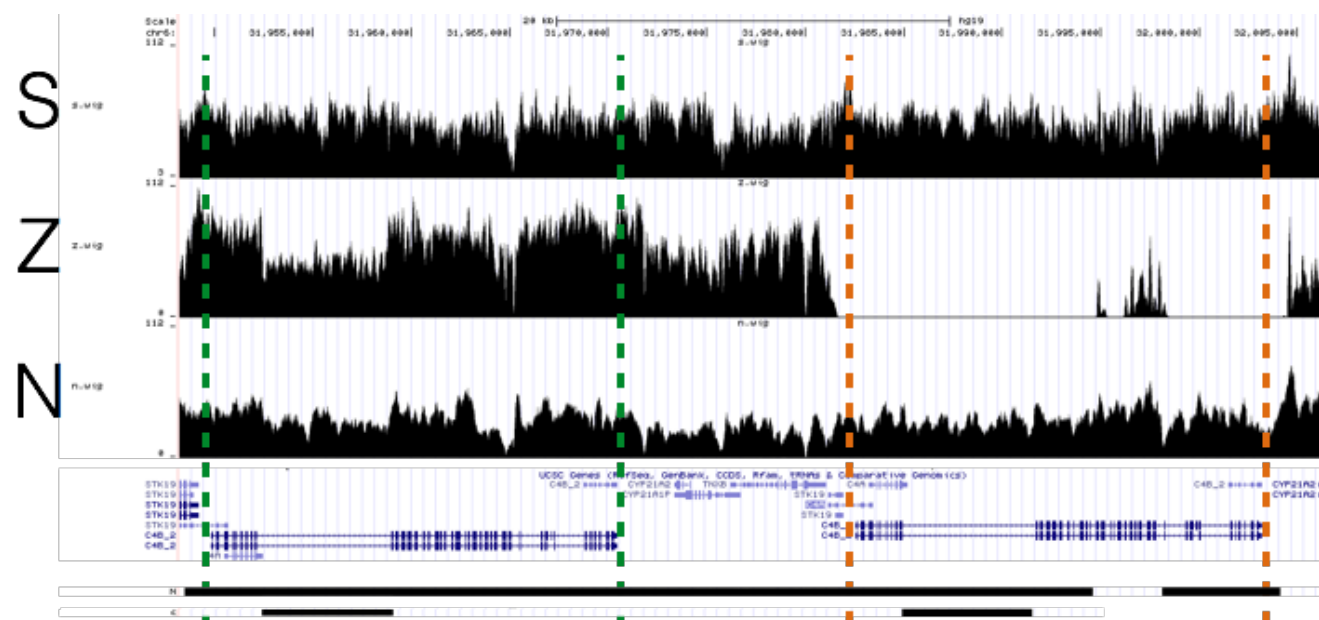
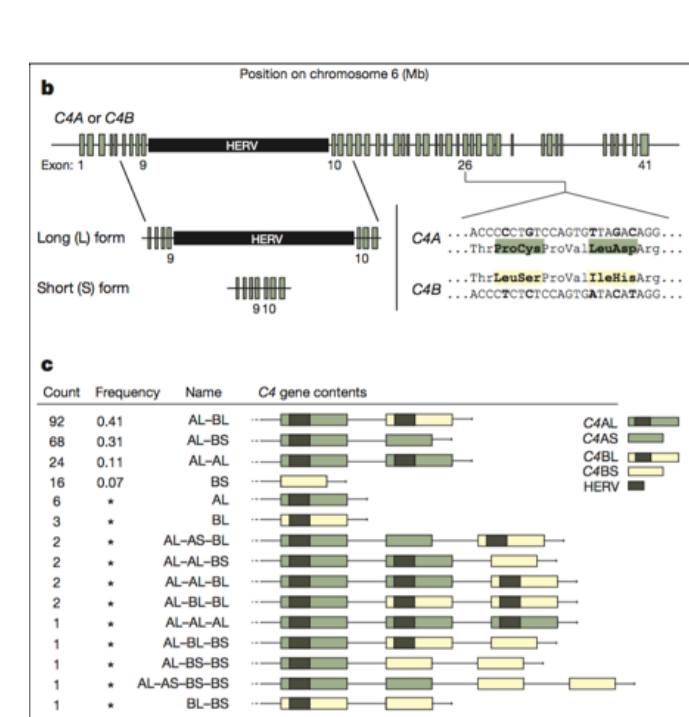


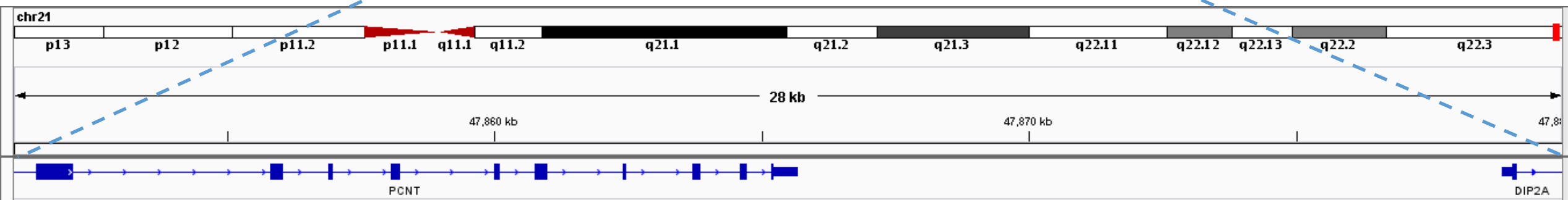
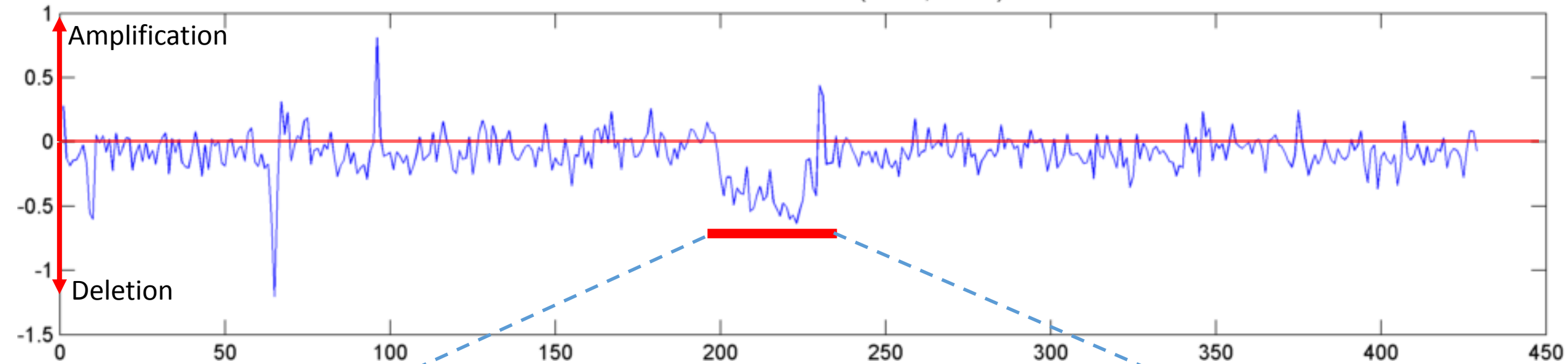
## Duplications



# Schizophrenia risk from complex variation of complement component 4

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



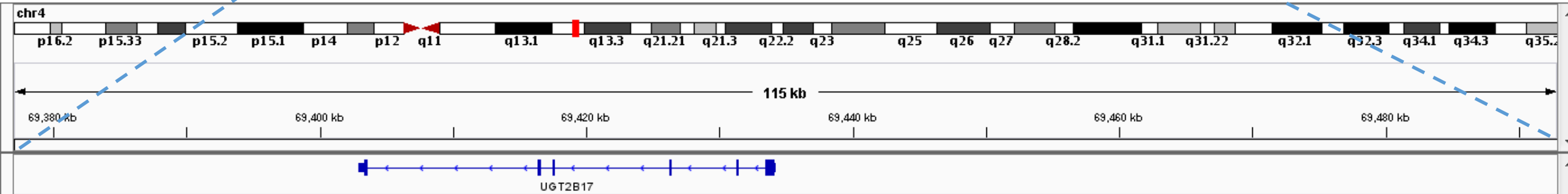
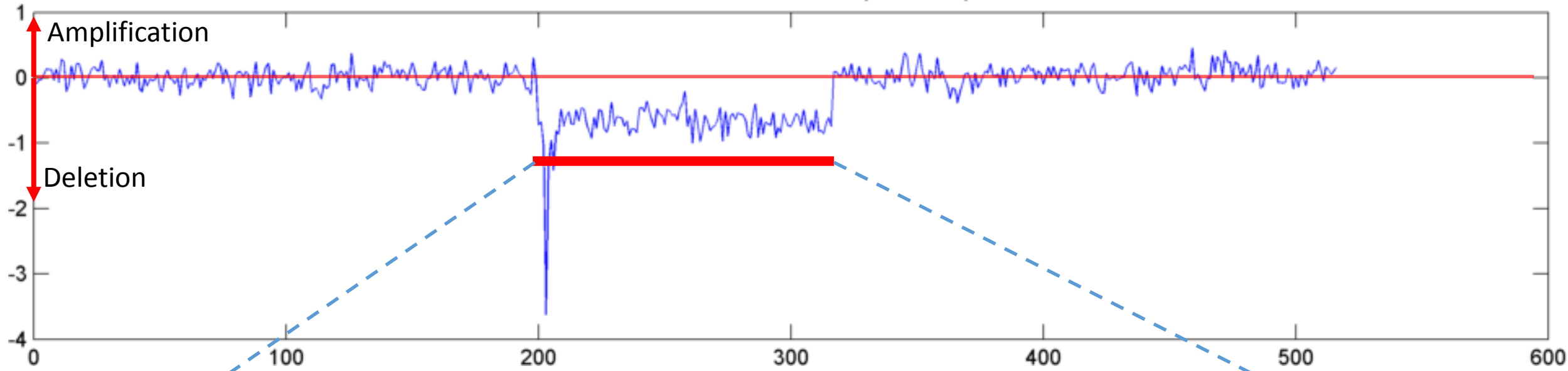


## Identification of Novel Dyslexia Candidate Genes Through the Analysis of a Chromosomal Deletion

G. Poelmans,<sup>1,2\*</sup> J.J.M. Engelen,<sup>1,3</sup> J. Van Lent-Albrechts,<sup>1</sup> B. Franke,<sup>4,5</sup> J.K. Buitelaar,<sup>5</sup> M. Wuisman-Frerker,<sup>6</sup> W. Eerenberg,<sup>1,3</sup> and C. Schrandt-Stumpel<sup>1,3</sup>

In 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

chr4:69377002-69493001 (UGT2B17)

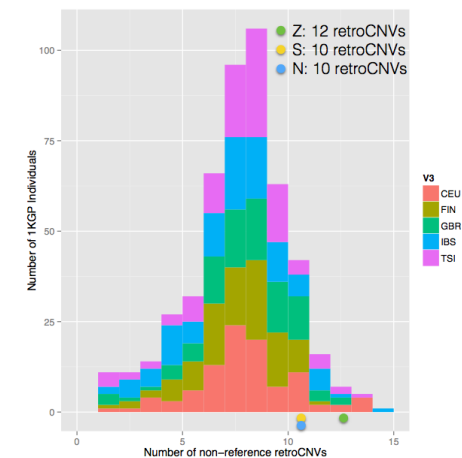
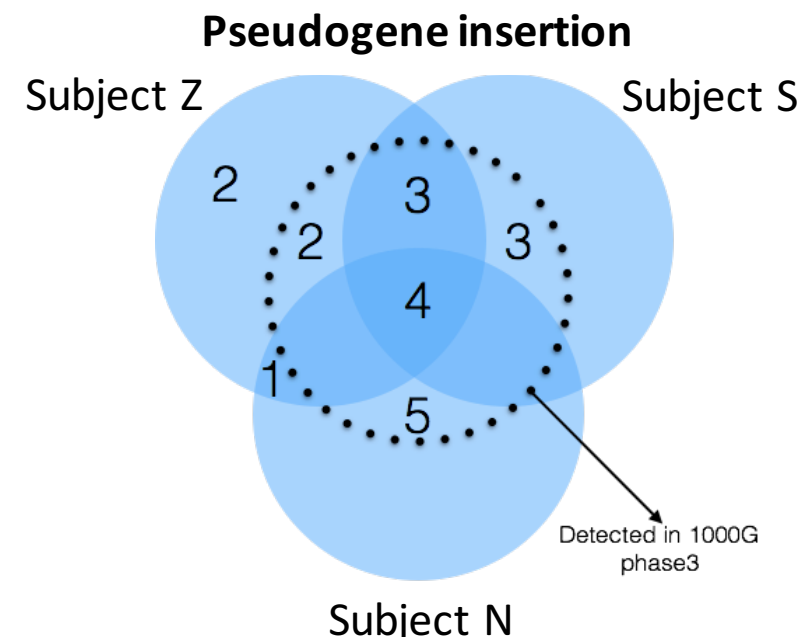
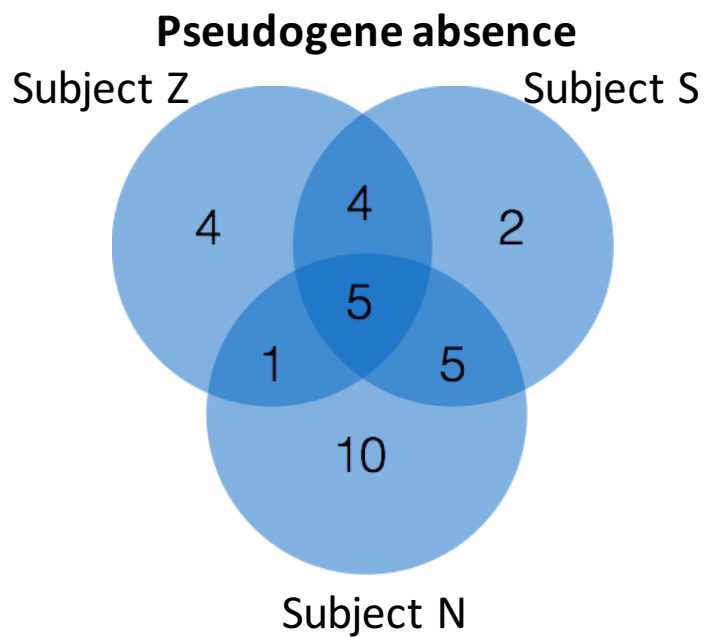
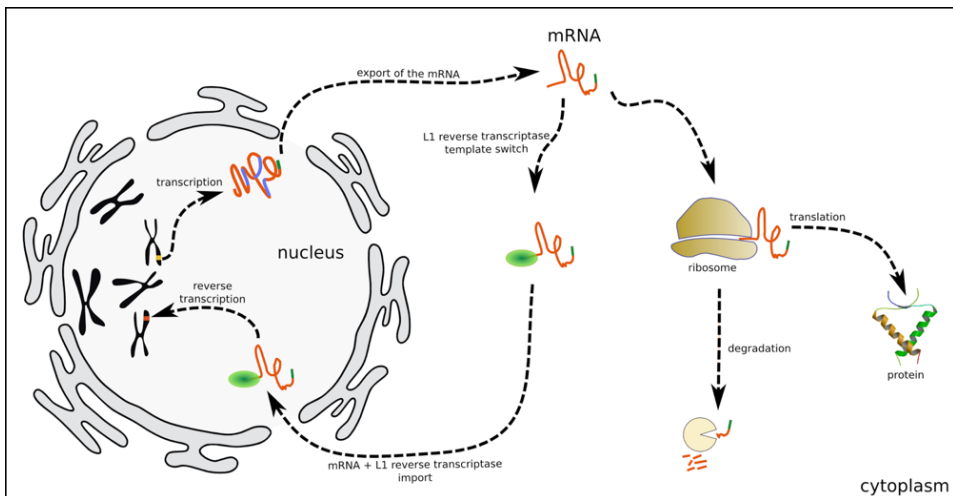


## Deletion Polymorphism of UDP-Glucuronosyltransferase 2B17 and Risk of Prostate Cancer in African American and Caucasian Men

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

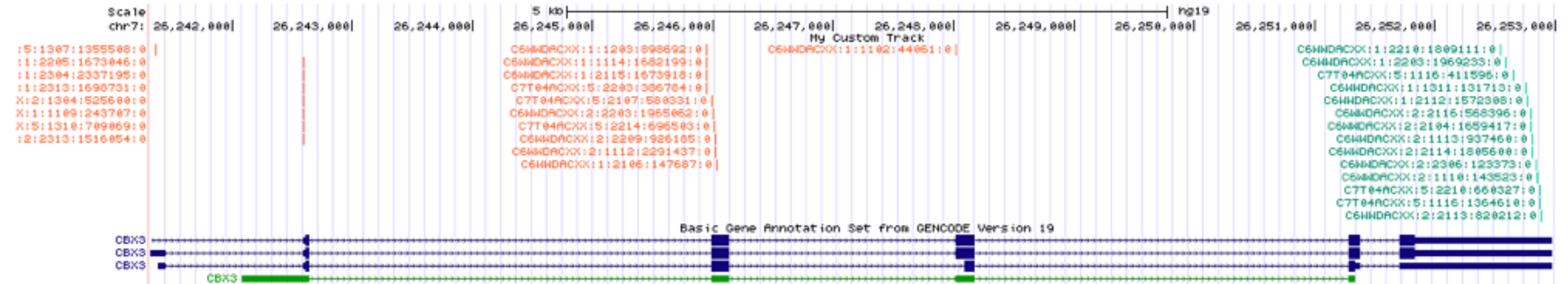
	Pseudogenes	Processed pseudogenes	Human specific processed pseudogenes
Human	~14,000	7,831	127



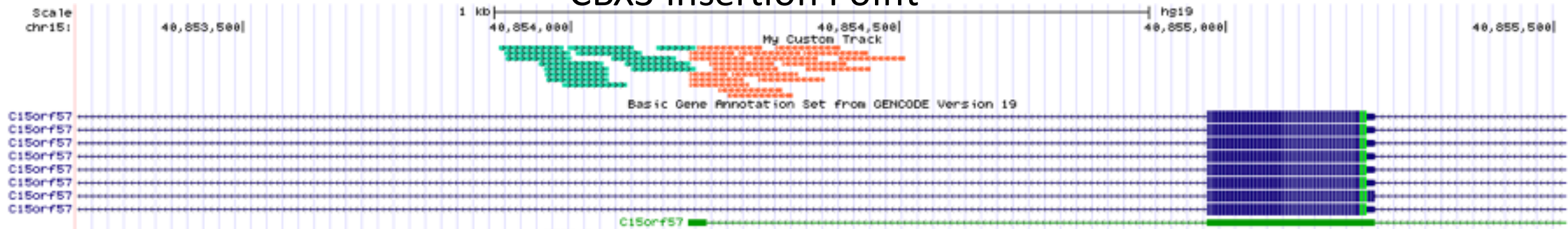


# Pseudogene CNV – Example I

## CBX3 Parental gene



## CBX3 Insertion Point





# ALU variation

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

