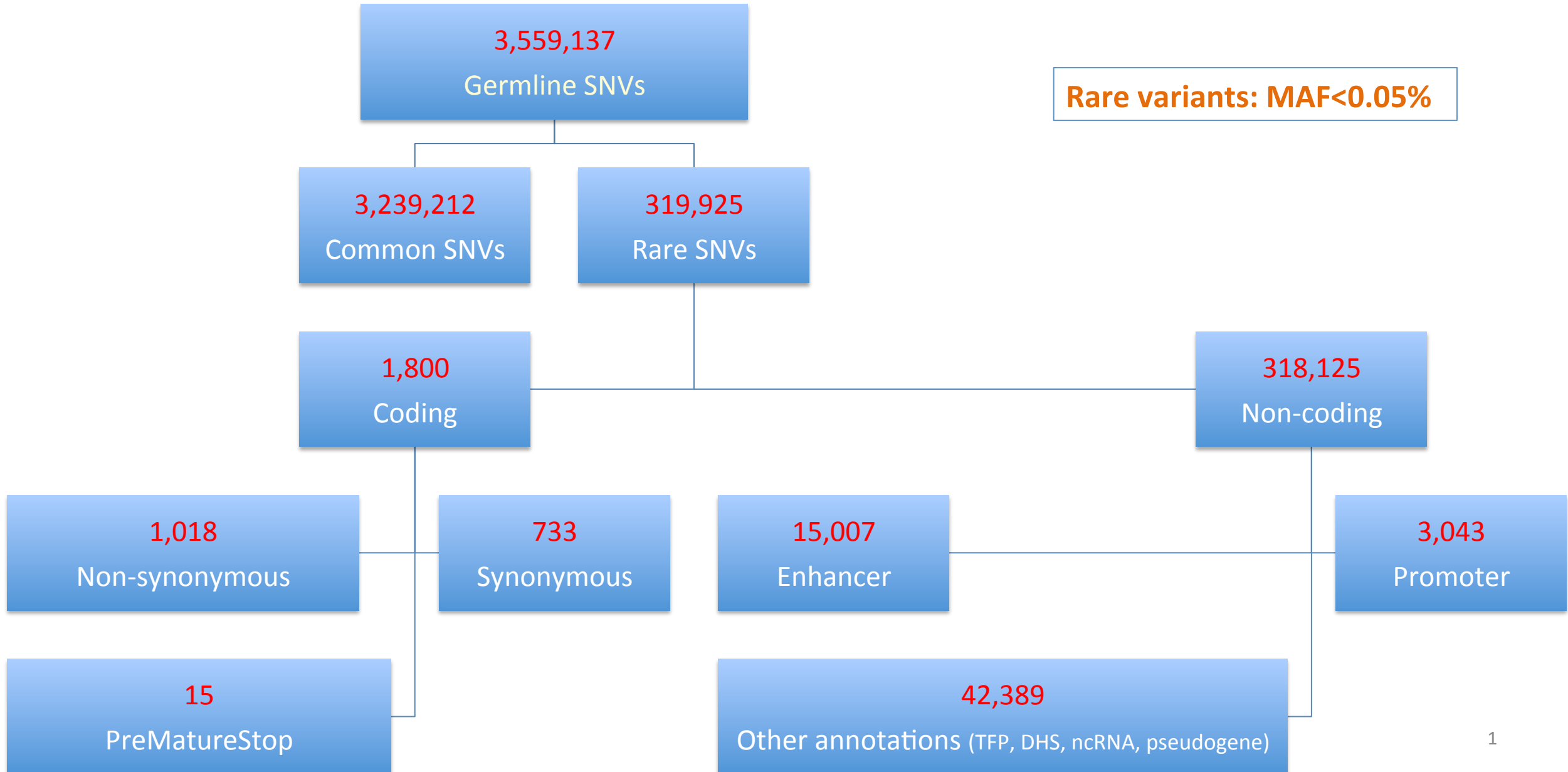
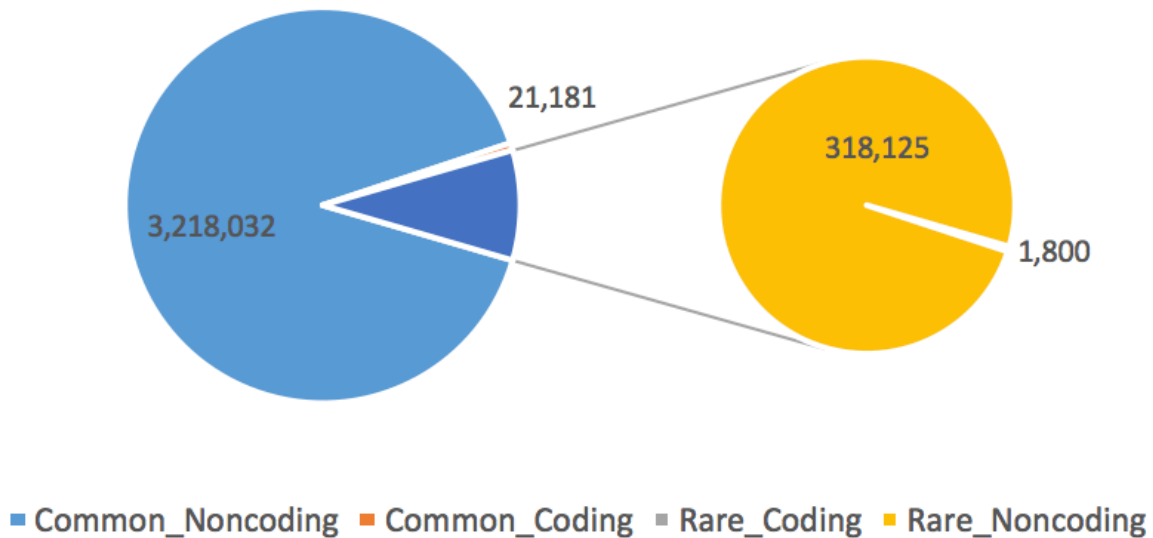


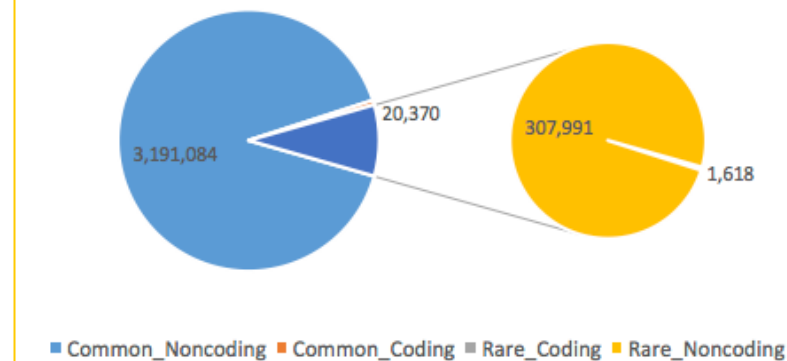
# Individual Z Overview



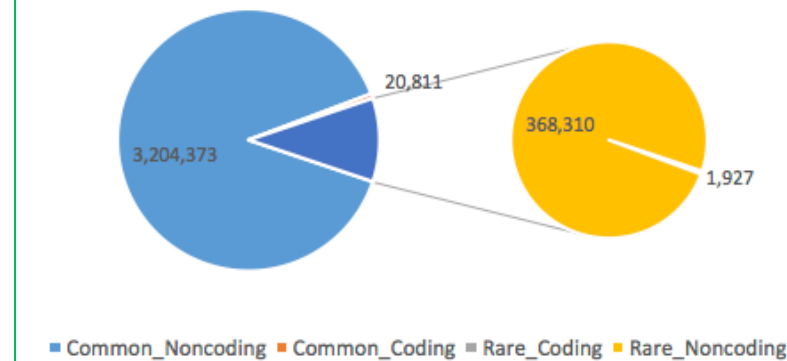
SNVs of Individual Z



SNVs of Individual S



SNVs of Individual NA



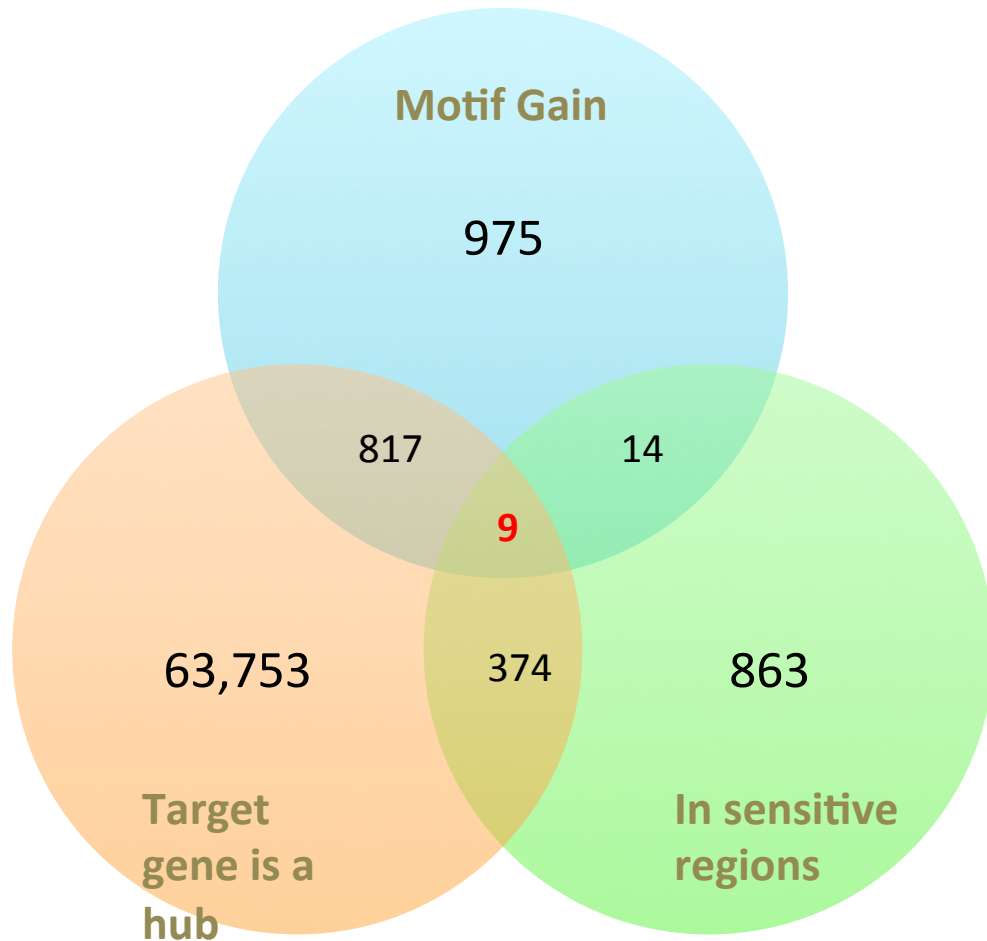
### Annotations for rare noncoding variants

	Subject Z	Subject S	Subject N
Promoter	3,043	2,473	3,038
Enhancer	15,007	14,186	15,190
Other noncoding annotation*	42,389	40,048	44,510

\* TFP, DHS, ncRNA, pseudogene

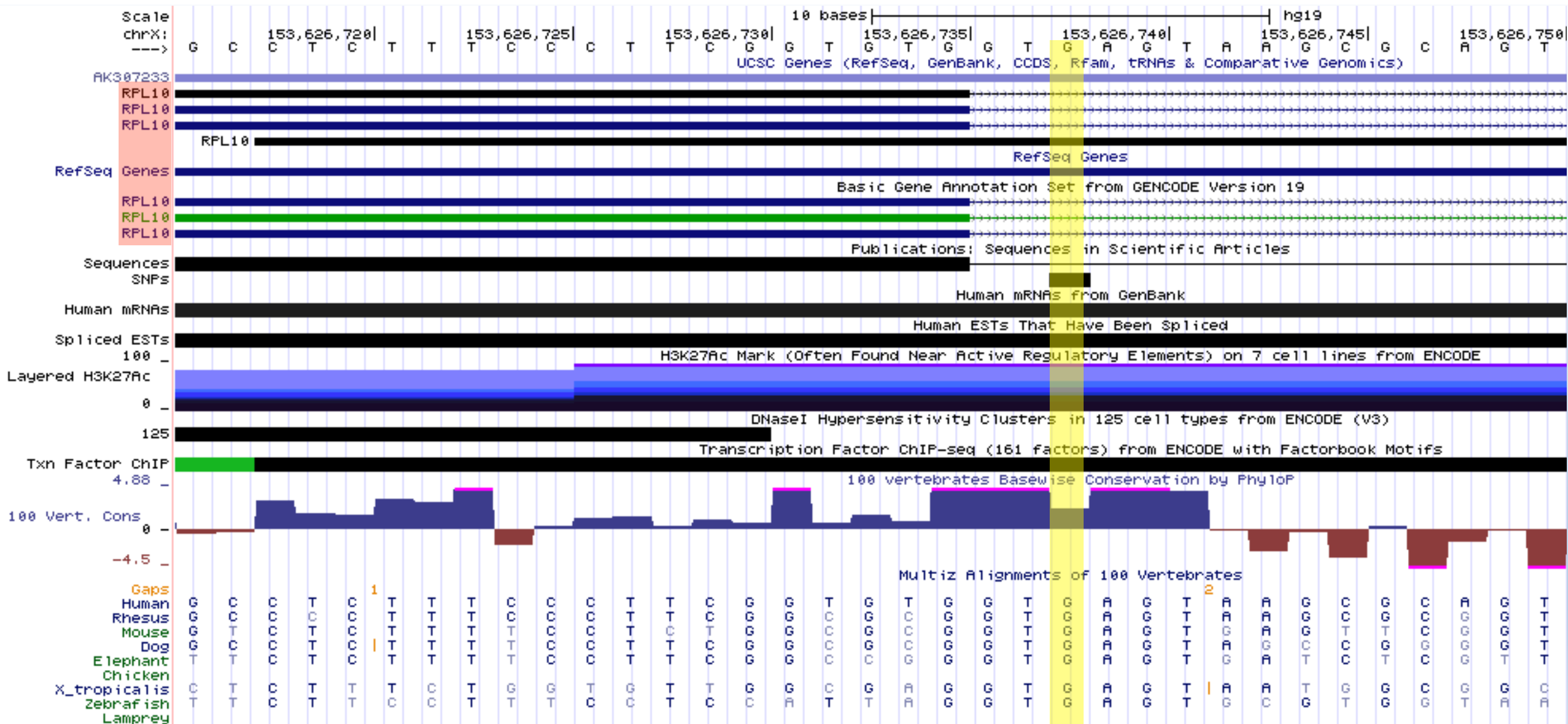
# Annotation of Rare Noncoding Variants

- 9 variants -> **11** target genes

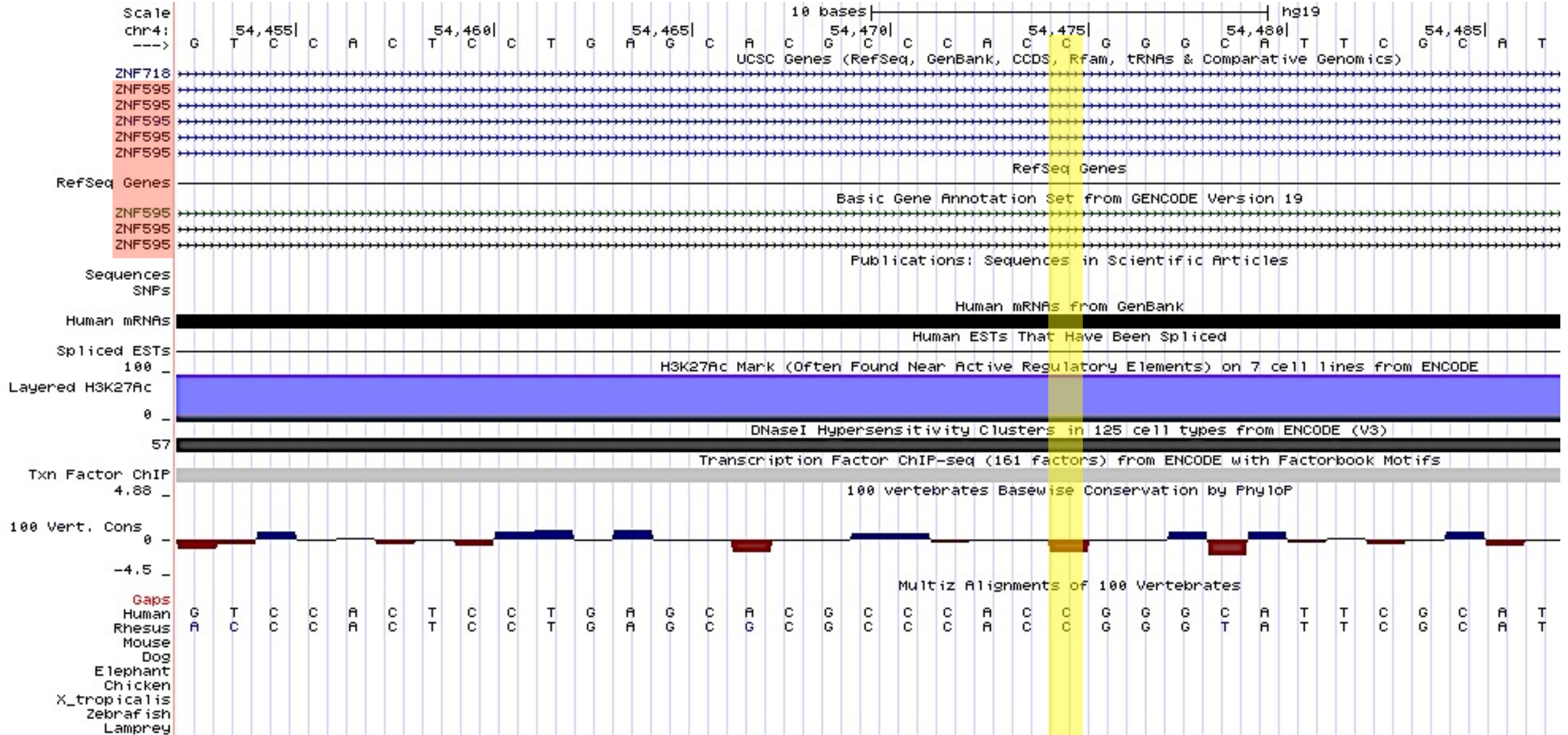


Gene Name	Variant Location	Function Annotation
RPL10	(Promoter&UTR)	[cancer]
PDE4DIP	(Distal&Intron)	[cancer]
ZNF595	(Intron&Promoter)	
GADD45G	(Promoter)	[DNA_repair]
CCND2	(Distal)	[actionable][cancer]
ACAP3	(Intron)	
VANGL2	(Promoter)	
SEC22B	(Distal)	
RNU1-9	(Distal)	
PARP11	(Distal)	
PUSL1	(Promoter)	

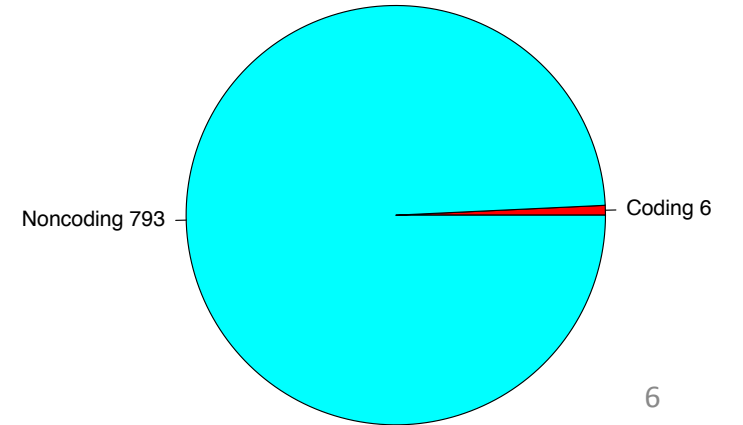
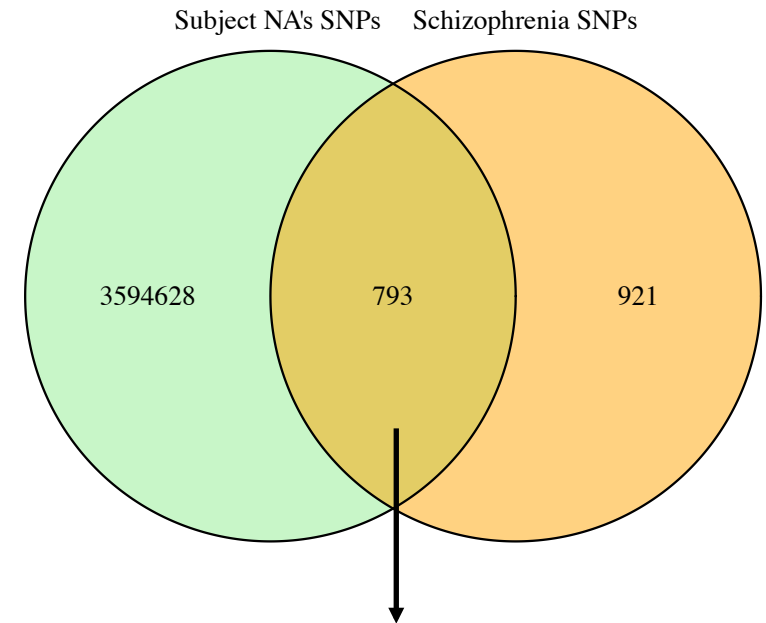
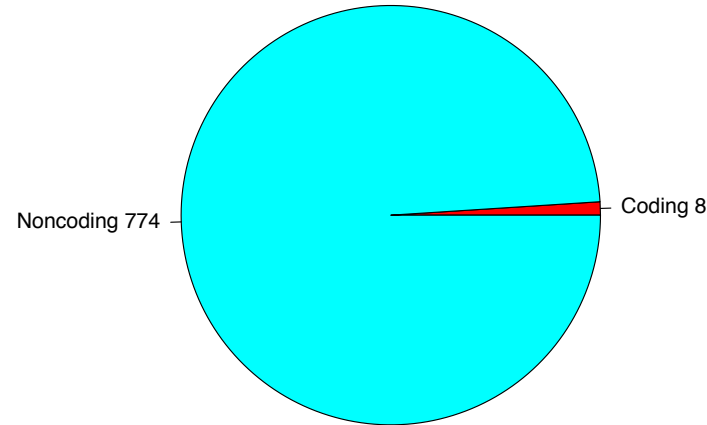
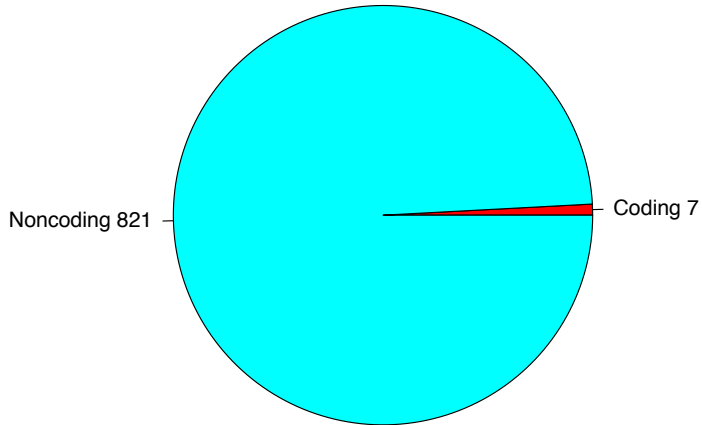
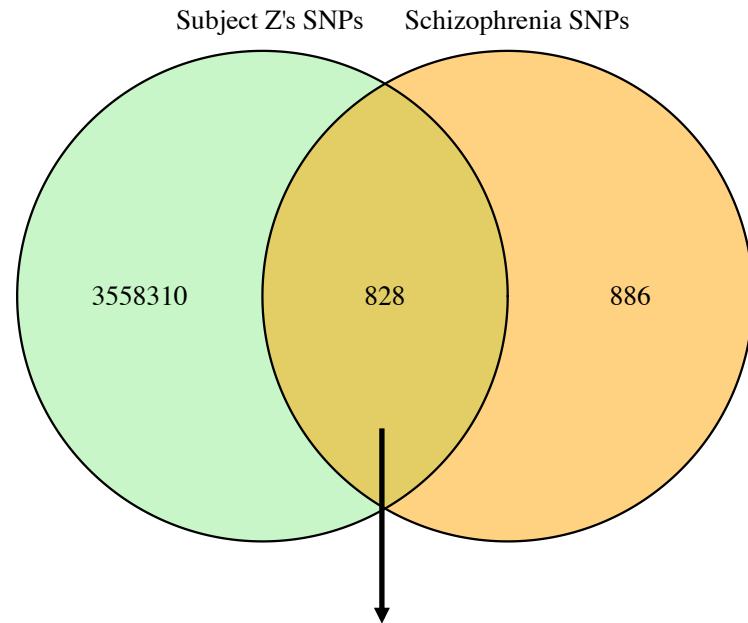
Chromosome	Position	Ref	Alt	Annotation	Target Gene
chrX	153626738	G	A	UTR & Promoter	RPL10 (ribosomal protein; autism & cancer)



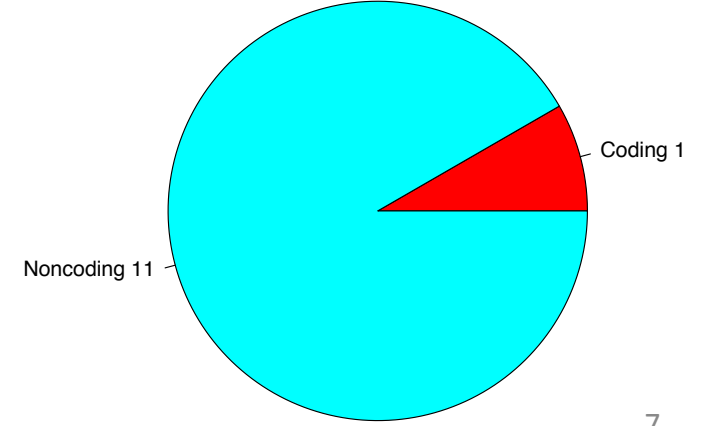
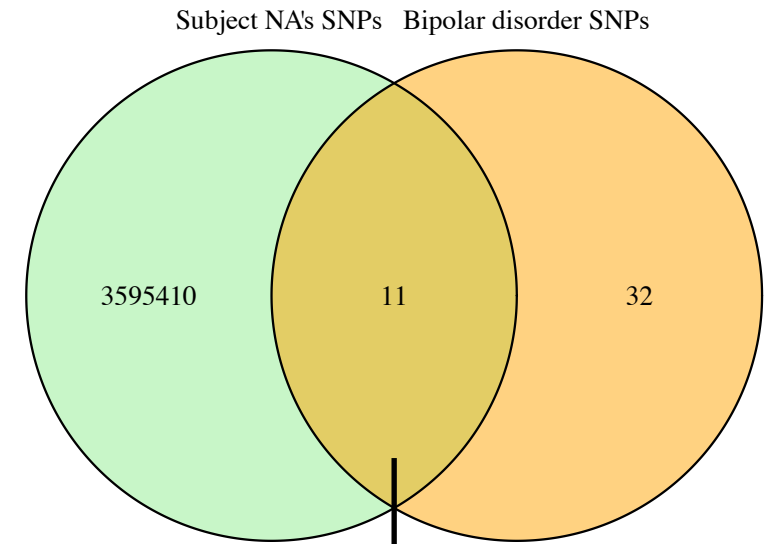
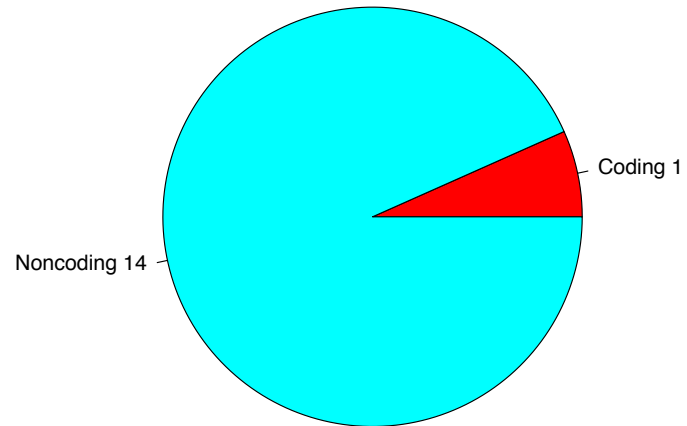
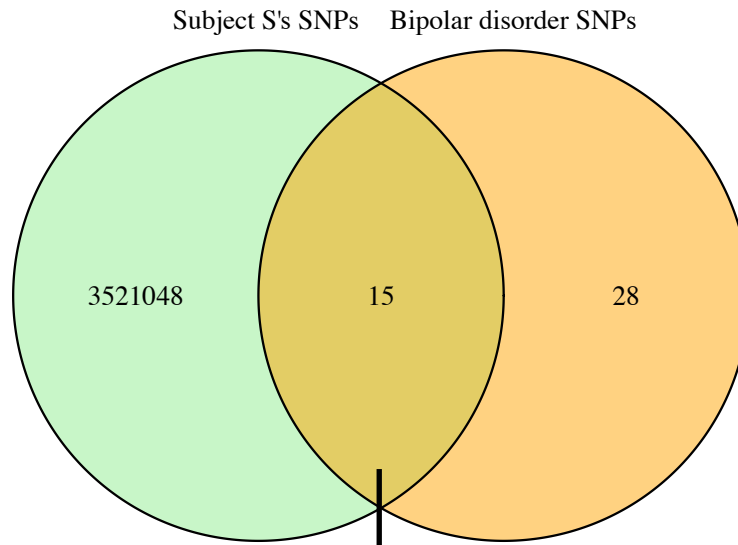
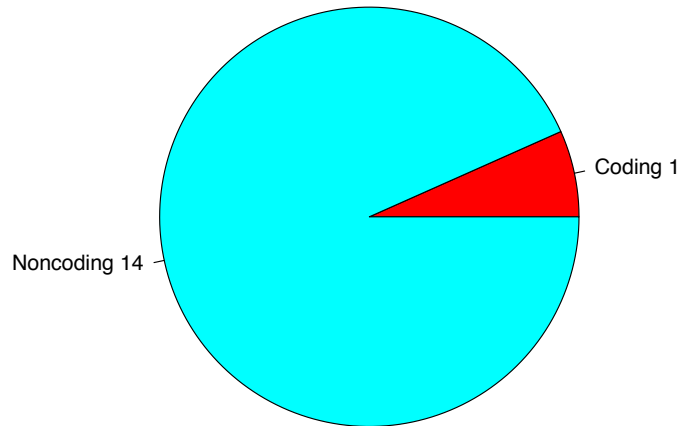
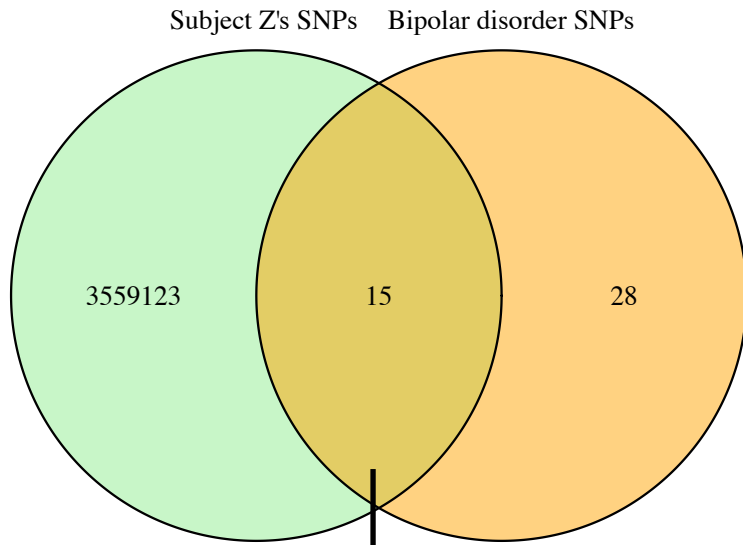
Chromosome	Position	Ref	Alt	Annotation	Target Gene
chr4	54475	C	T	Intron & Promoter	ZNF595 (zinc finger protein; gene expression)



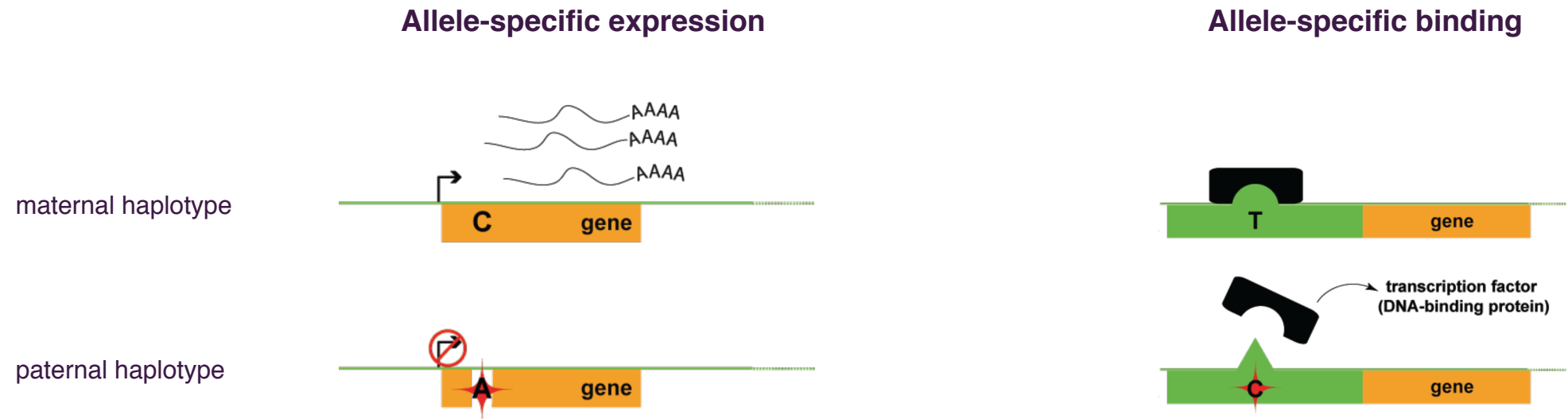
# Overlap with 1714 Schizophrenia SNPs



# Overlap with 43 Bipolar disorder SNPs



# Subject Z's SNVs associated with allele-specific expression (ASE) and binding (ASB)



- Subject Z has **2,081,803** heterozygous SNVs
- **40,267** and **19,687** of them were found in AlleleDB, assessed for ASE and ASB, respectively
- **8,680** (ASE) and **734** (ASB) SNVs show some evidence for allele-specific behavior: allele-specific in at least one of the 380 AlleleDB individuals
- **173** of SNVs show consistent association with ASE across AlleleDB individuals: present in more than 100 individuals and found to be allele-specific in more than 95% of them



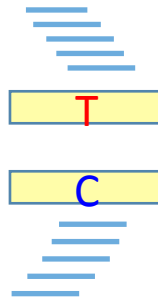


# Subject Z's SNVs associated with allele-specific expression (ASE) and binding (ASB)

## Expression/binding balanced between alleles

### RNA-/ChIP-Seq Reads

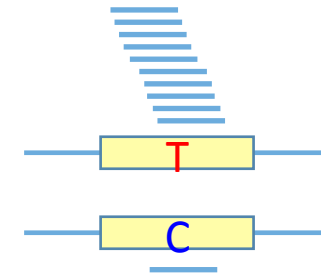
ACTTTGATAGCGTCAAC**CG**  
 CTTTGATAGCGTCAAC**CGC**  
 CTTTGATAGCGTCAAC**CGC**  
 TTGACAGCGTCAAT**TGCAC**  
 TGATAGCGTCAAT**TGCACG**  
 ATAGCGTCAAC**CGC**ACGTC  
 TAGCGTCAAT**TGCACGTCG**  
 CGTCAAC**CGC**ACGTCGGGA  
 GTCAAT**TGCACGTCGAGAG**  
 CAAT**TGCACGTCGGGAGTT**



## Allele-specific expression/binding

### RNA-/ChIP-Seq Reads

ACTTTGATAGCGTCAAT**TG**  
 CTTTGATAGCGTCAAT**TGC**  
 CTTTGATAGCGTCAAT**TGC**  
 TTGACAGCGTCAAT**TGCAC**  
 TGATAGCGTCAAT**TGCACG**  
 ATAGCGTCAAT**TGCACGTCG**  
 TAGCGTCAAT**TGCACGTCG**  
 CGTCAAC**CGC**ACGTCGGGA  
 GTCAAT**TGCACGTCGAGAG**  
 CAAT**TGCACGTCGGGAGTT**



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