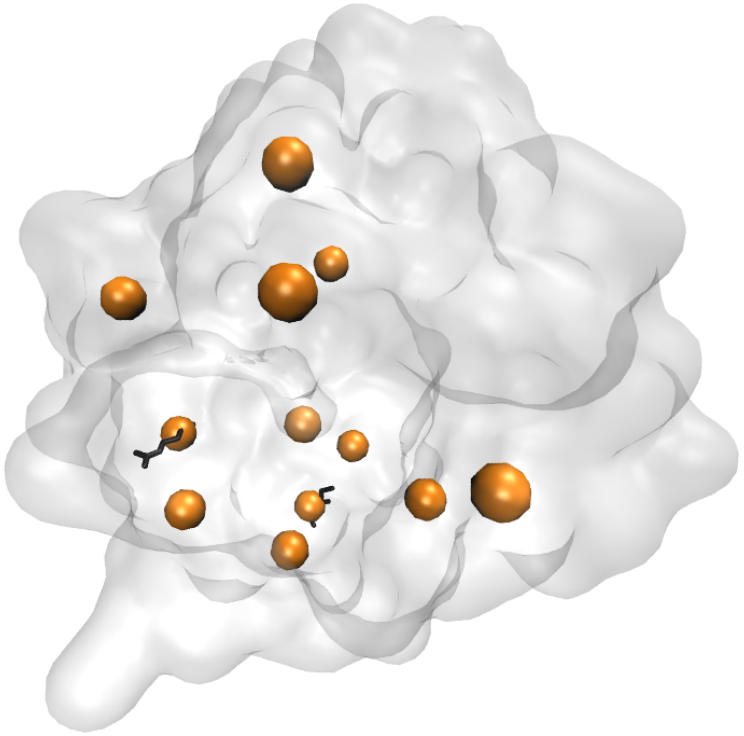
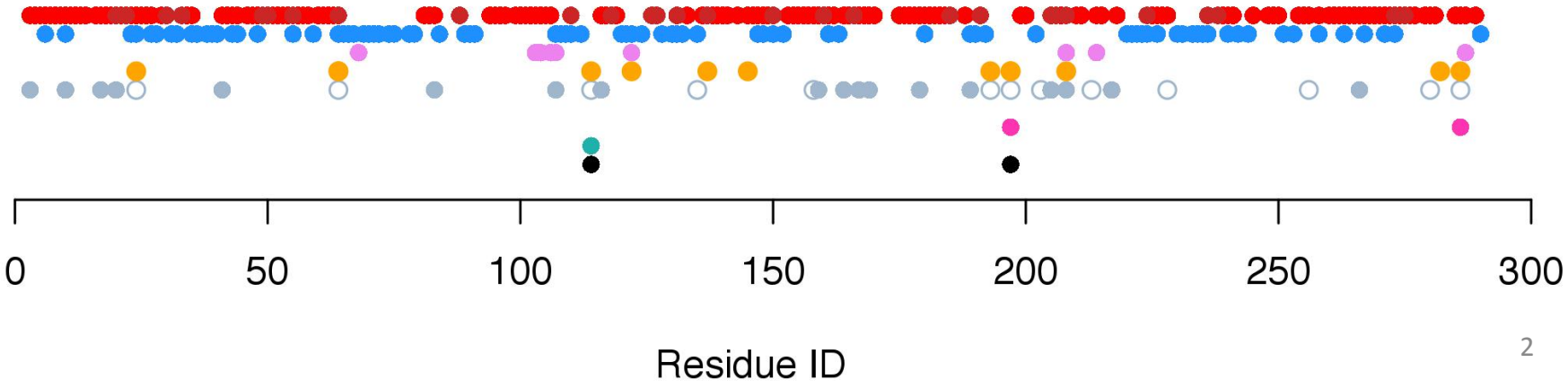
Coding Variation in Subject Z

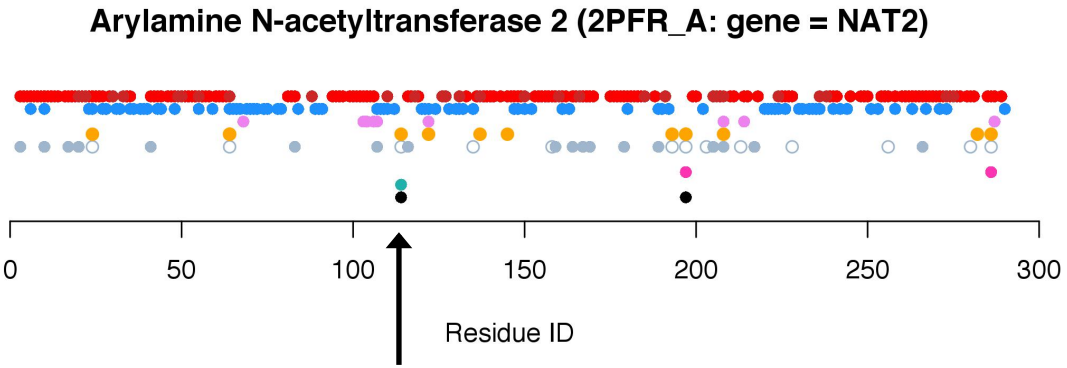
AlleleDB, ALOFT, STRESS



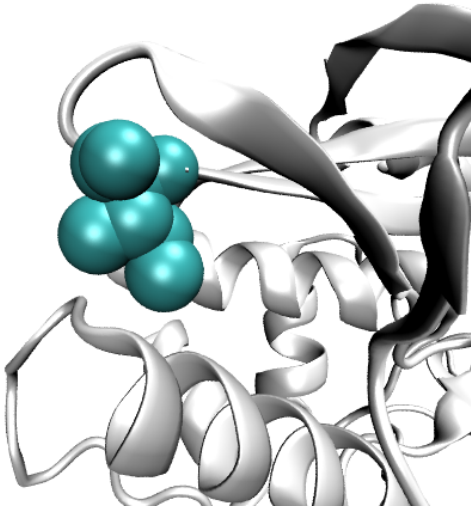
- Predicted allosteric (surface | interior)
- Buried residues
- Protein-protein interaction site
- Post-translational modifications
- HGMD (prem. stop | non-synon)
- 1000 Genomes (rare | common)
- Snyder (prem. stop | non-synon)
- na12878 (prem. stop | non-synon)
- Subj. Z (prem. stop | non-synon)

Arylamine N-acetyltransferase 2 (2PFR_A: gene = NAT2)

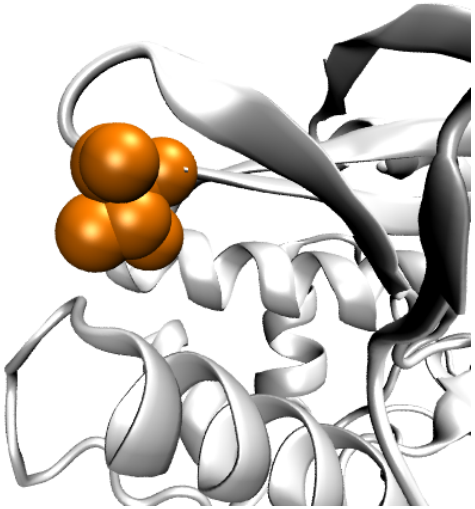




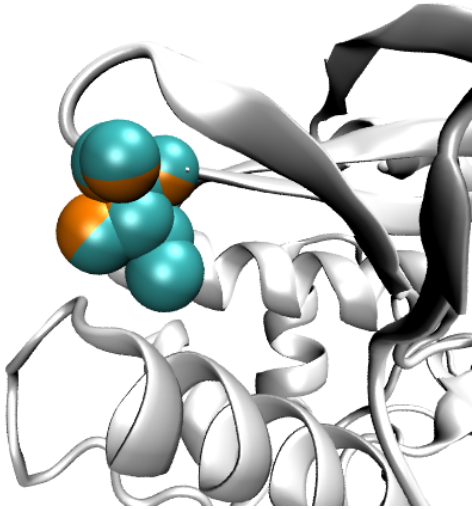
114: I->T



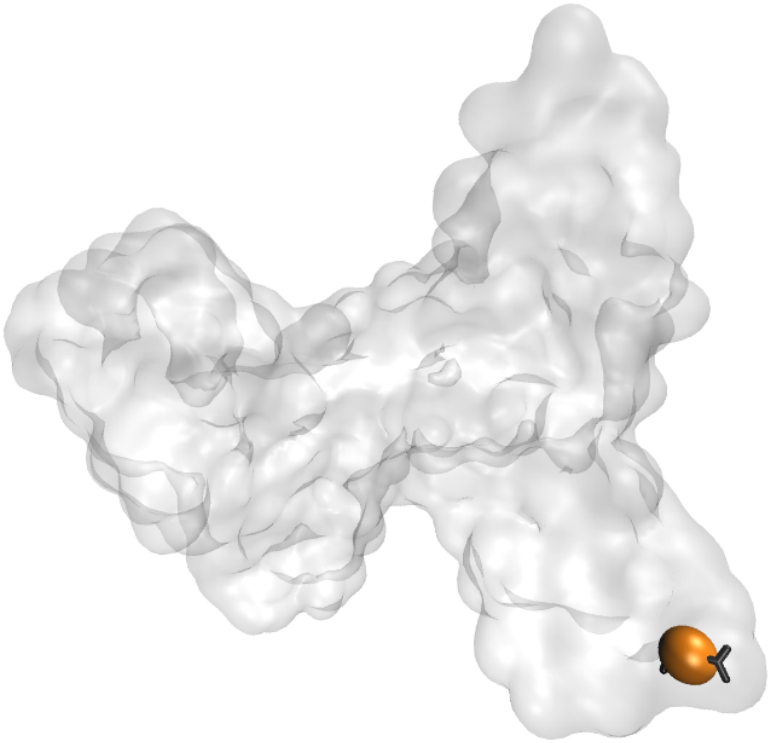
Wild-type



Mutated



(superimposed)



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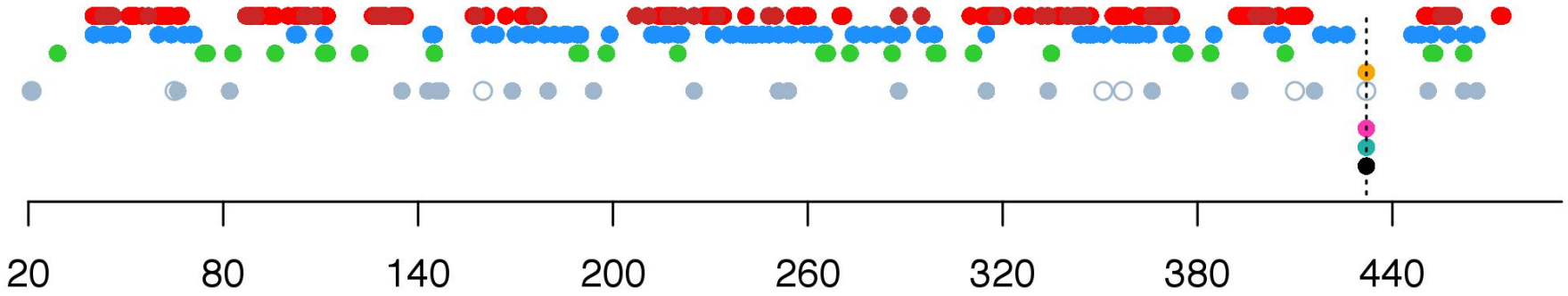
Vitamin D-binding protein (1KW2_A: gene = GC)



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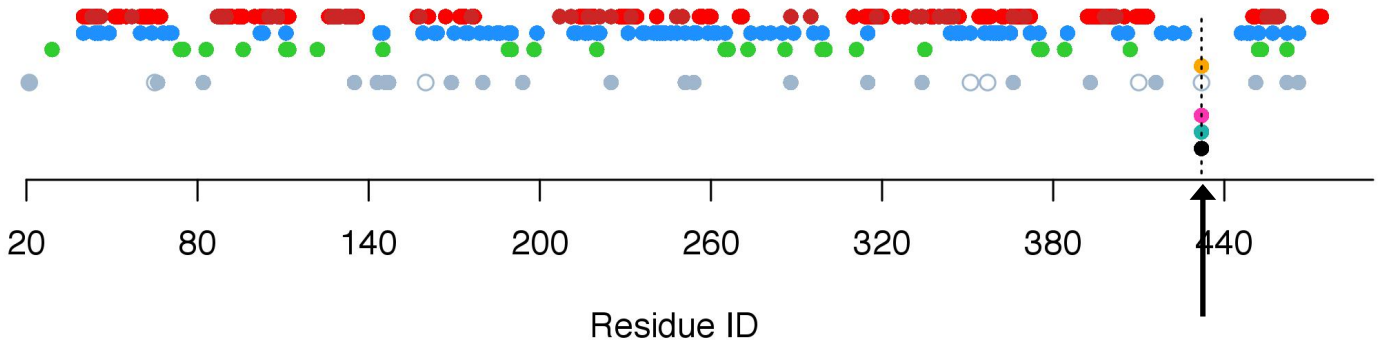
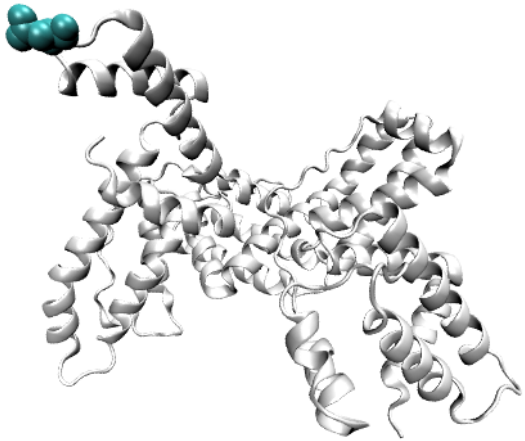
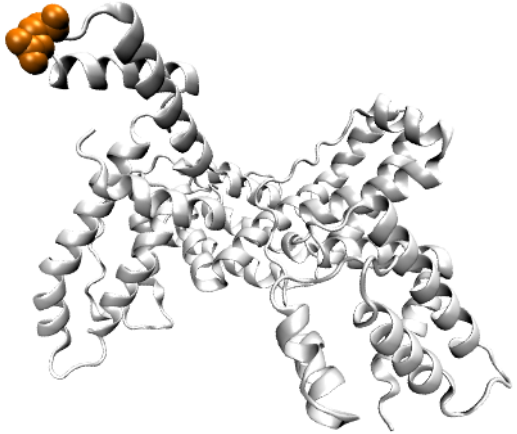


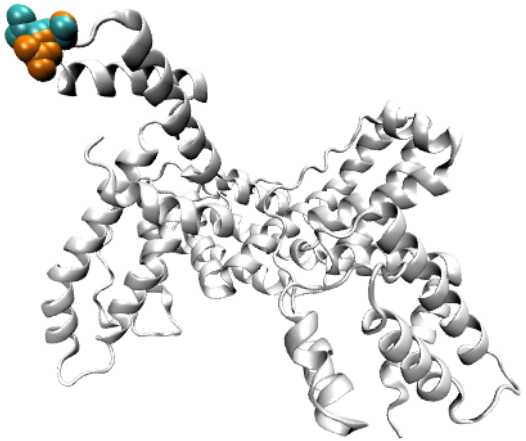
432 D->E



Wild-type

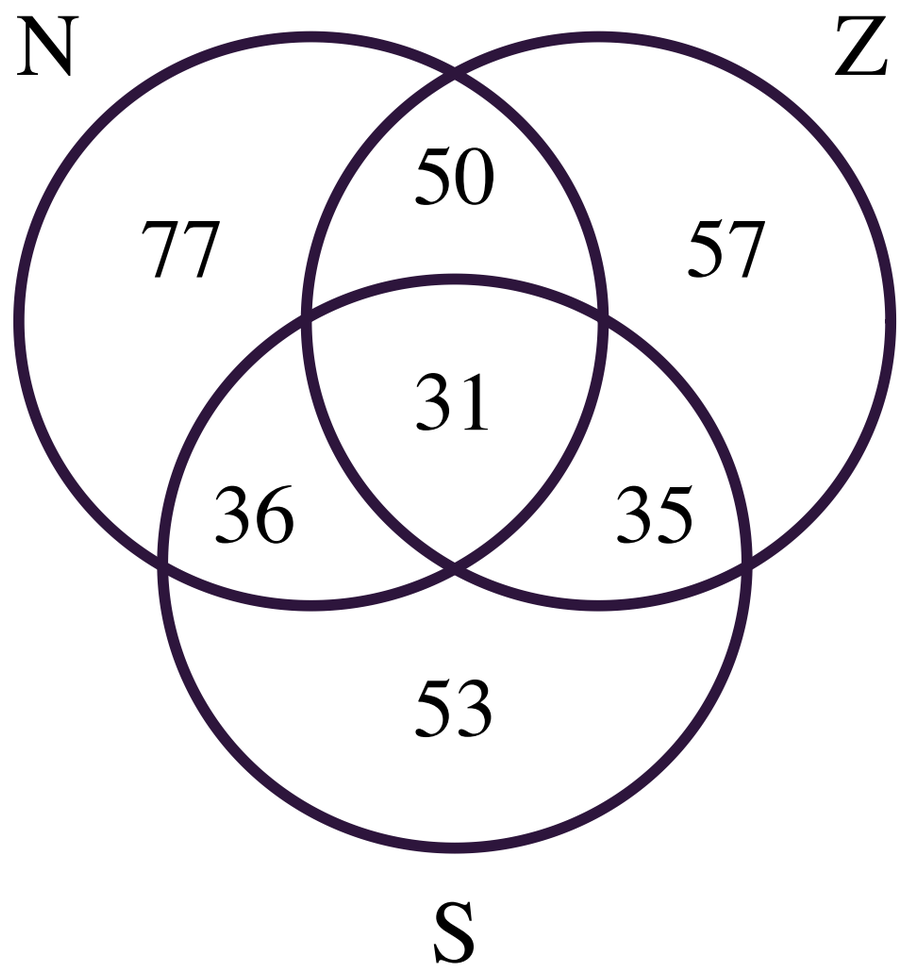


Mutated



(superimposed)

40,078 (/3,595,421) *N,* **38,999** (/3,521,063) *S*, and **40,304** (/3,559,138) *Z* HetSNPs in AlleleDB



Number of SNVs which are present in >100 AlleleDB individuals and are associated with allele-specific expression in more than 95% of them

XL PIE CHARTS

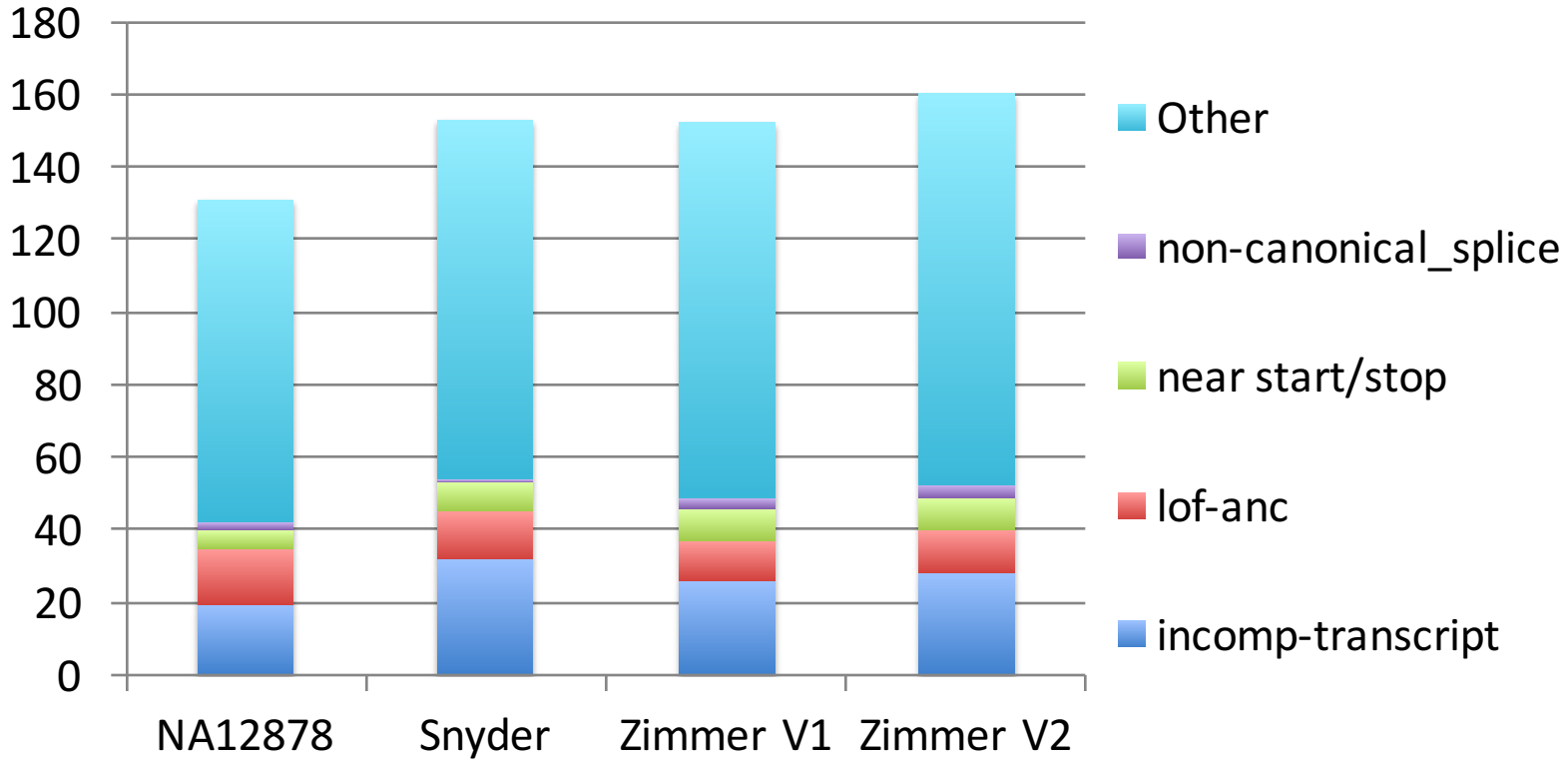
Rare Non-synonymous Coding Variants

- 1018 SNVs -> 824 target genes

Gene Annotation	Gene Name
Cancer-related	NOTCH2; PDE4DIP; TPR; CRTC3; CDH11; MLLT6; ASXL1; HMGA1; KDM6A
DNA repair	RECQL; RAD51; PPM1D; XRCC1; AP1B1; FANCI; PTPRH; RBBP7; SLX4; POLR2A; DCLRE1C; ANKLE1
Cancer & DNA repair	ATM; PMS2; ERCC5
Actionable Gene	ATM; KDM6A; INSR; FOXP4

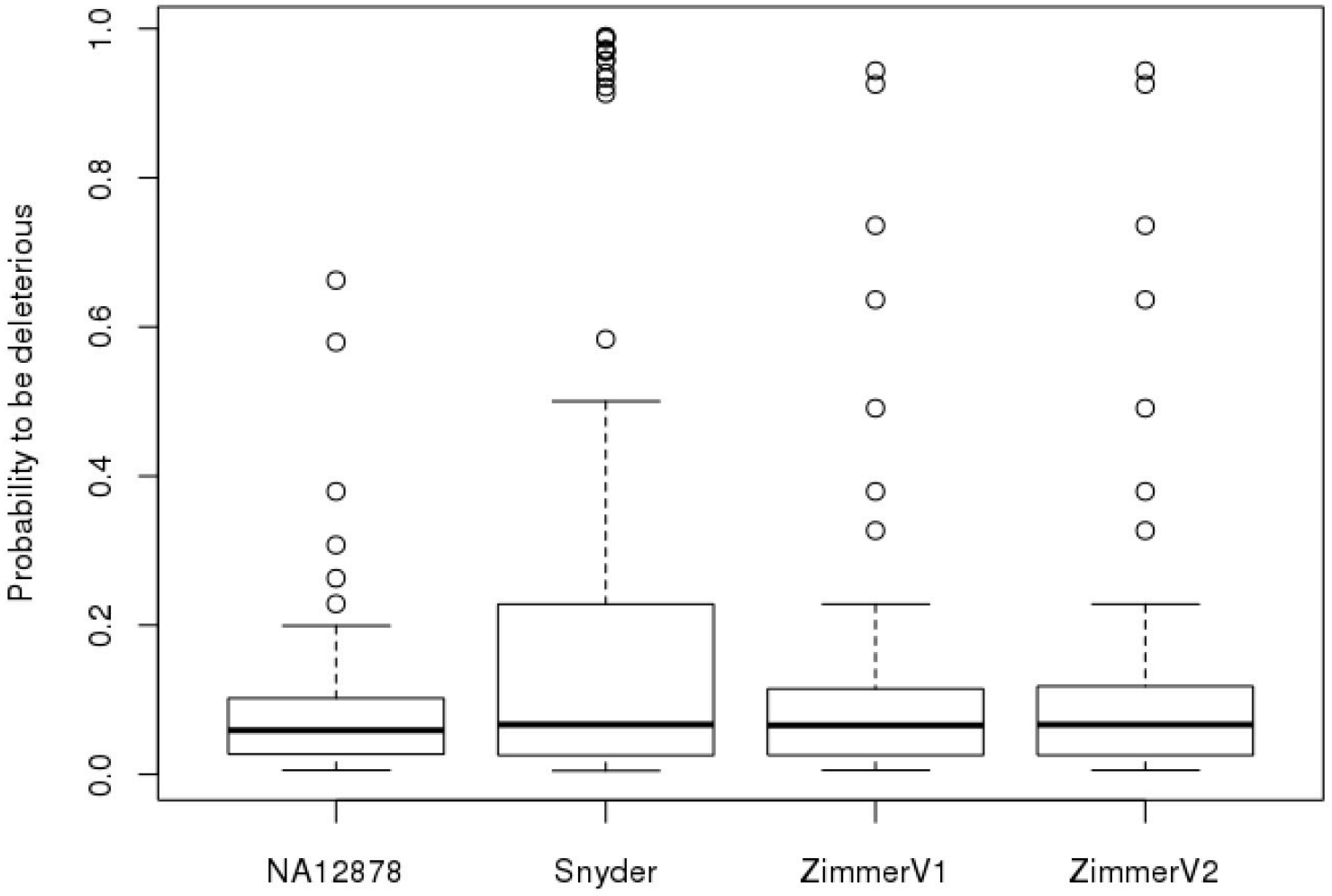
- ATM: Serine/Threonine Kinase; Regulator of p53 and BRCA1; leukemia; ataxia-telangiectasia; breast cancer
- PMS2: Direct p53 effectors; mismatch repair cancer syndrome; colorectal cancer; hereditary nonpolyposis
- ERCC5: Chks in Checkpoint Regulation; DNA Repair; xeroderma pigmentosum
- KDM6A: Transcriptional misregulation in cancer
- INSR: Insulin Receptor; PI3K-Akt signaling pathway; GPCR Pathway; Diabetes mellitus
- FOXP4: Transcriptional repressor that represses lung-specific expression

- **Transcript-level Flags**
- **LoF**



	Other
NA12878	89 (56 NMD)
Snyder	99 (67 NMD)
Zimmer V1	103 (58 NMD)
Zimmer V2	108 (63 NMD)

LOF variants



Potential deleterious variant
Zimmer

chr	pos	ref	alt	gene	Score	genotype	Gene function
6	17606162	C	T	FAM8A1	0.94365	0/1	Unknown, Autism related ??? Pubmed: 22495306
6	155577717	T	A	TIAM2	0.63655	0/1	Cell migration
17	61829719	A	C	CCDC47	0.92540	0/1	unknown
19	759925	C	A	MISP	0.73605	0/1	Mitotic spindle positioning

No disease associations in OMIM

CCDC47 associated with Schizophrenia