

N/S/Z 'omes:

SNVs associated with allele-specific expression

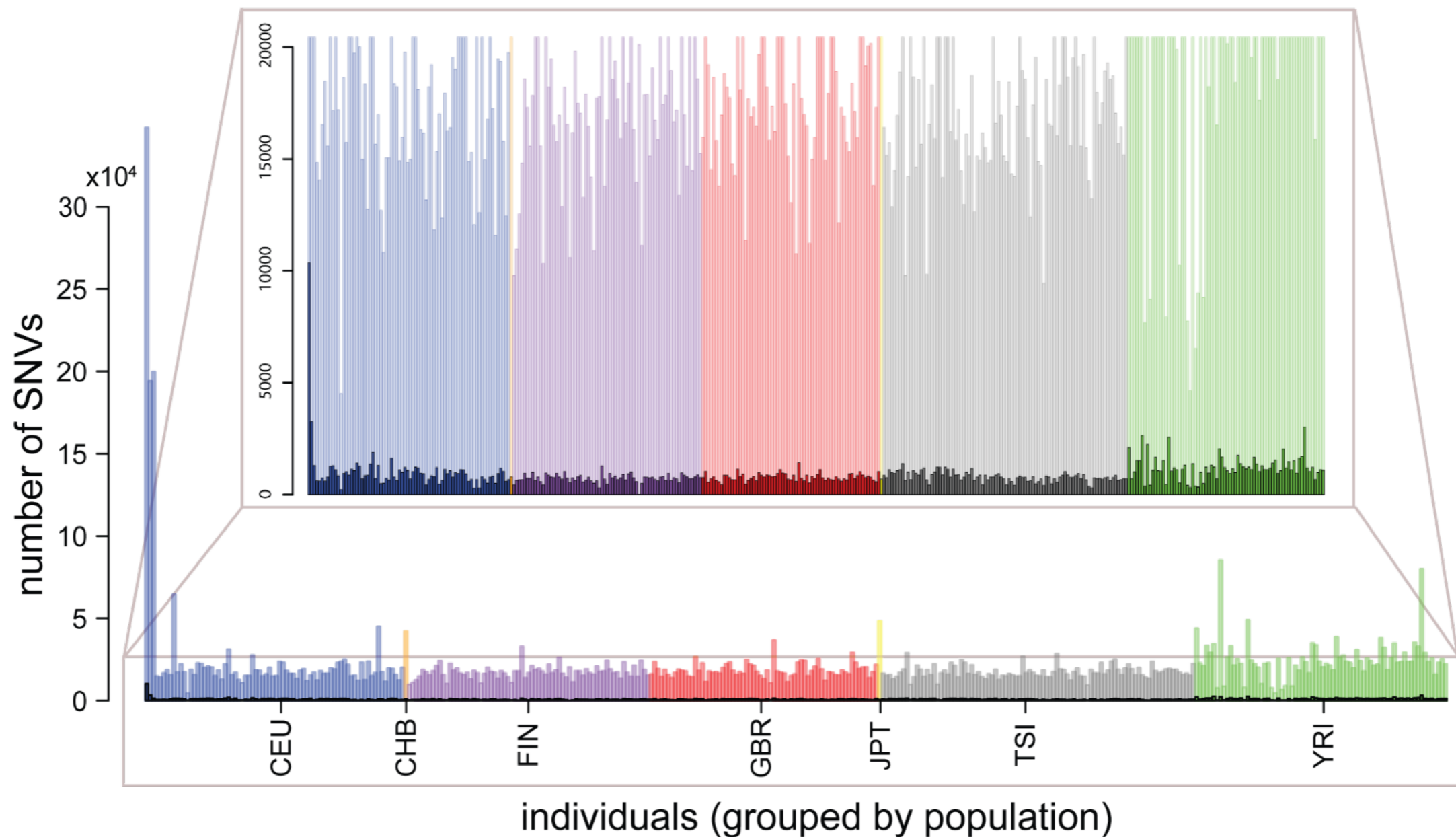
TG, JC, JR
Feb. 17, 2016

AlleleDB



alleledb.gersteinlab.org

632,126 ASE/nonASE-annotated SNVs across **383** individuals



342,500 ASE/nonASE-annotated SNVs across **380** individuals after removing the NA12878-NA12891-NA12892 trio

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

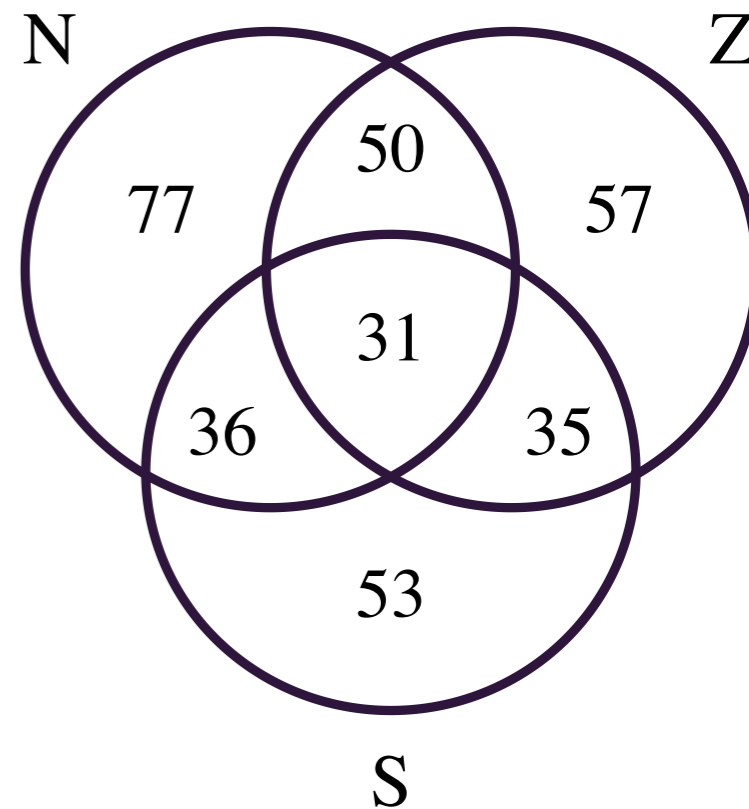
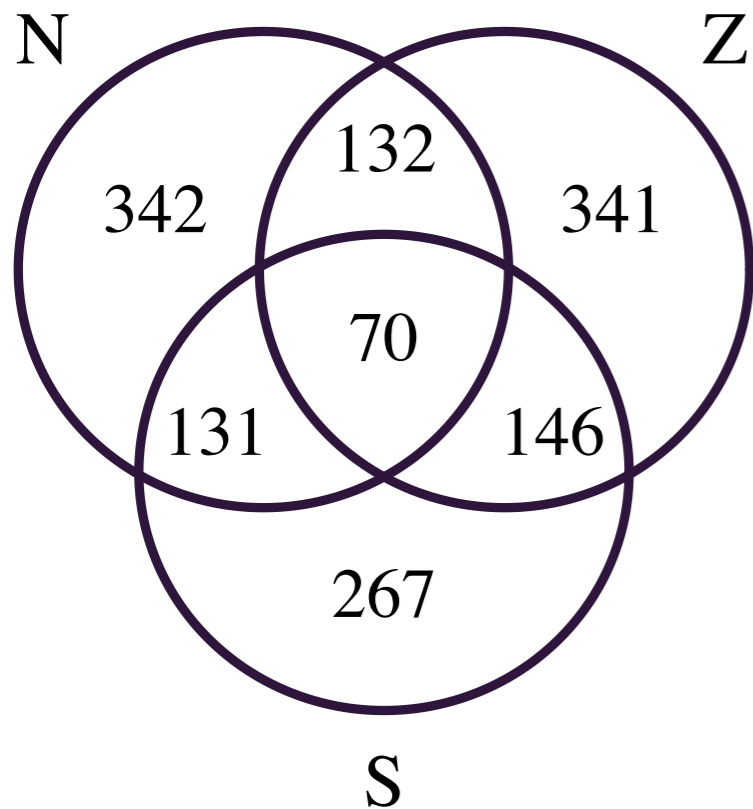
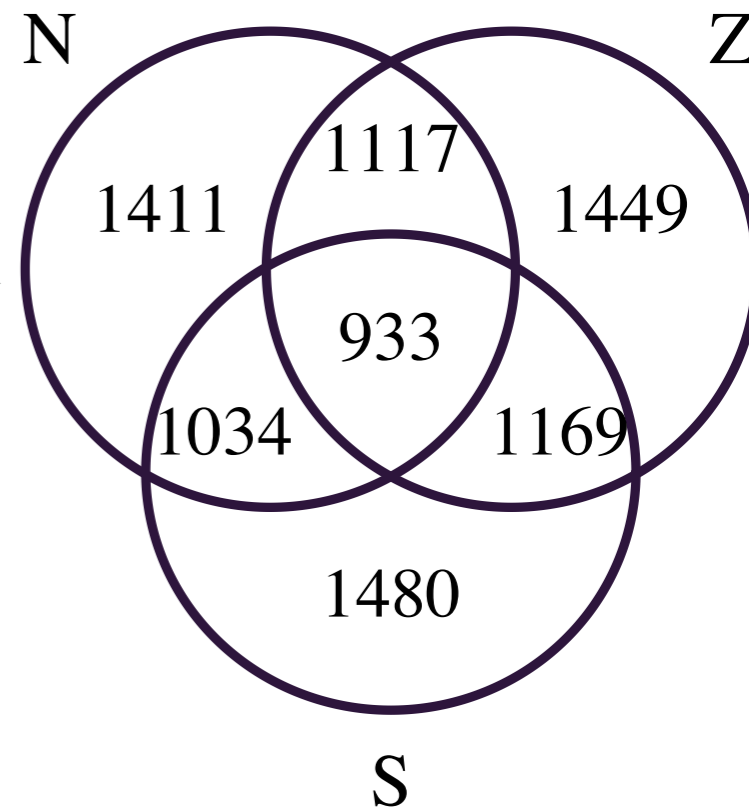
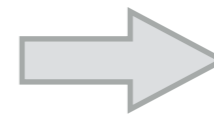
Number of N/S/Z HetSNPs matching HetSNPs in AlleleDB individuals

columns: total count of individuals with the HetSNP accessible,

rows: fraction of individuals in which the HetSNP is associated with ASE.

	1	2-4	5-19	20-99	100-380
[0.00, 0.05)	9956/9710/10149	7082/6835/6839	5566/5412/5624	7809/7819/8012	4495/4616/4668
[0.05, 0.25)	0/0/0	0/0/0	971/959/992	936/771/863	239/210/201
[0.25, 0.75)	0/0/0	602/554/613	356/324/376	306/259/300	82/68/72
[0.75, 0.95)	0/0/0	23/16/22	61/62/82	77/66/64	24/34/20
[0.95, 1.00]	675/614/689	245/221/250	133/119/126	246/175/169	194/155/173

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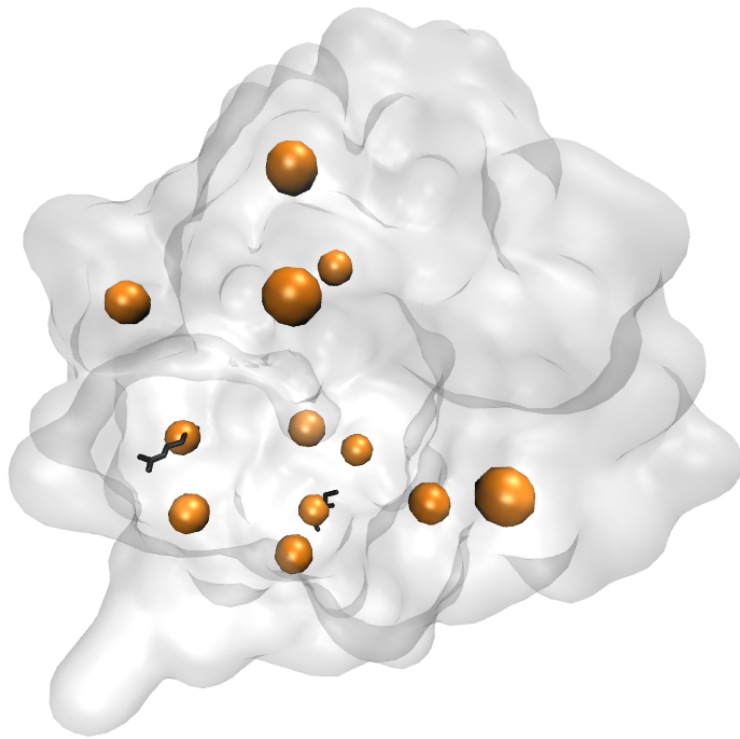


Rare Non-synonymous Coding Variants

- 1018 SNVs -> **824** target genes

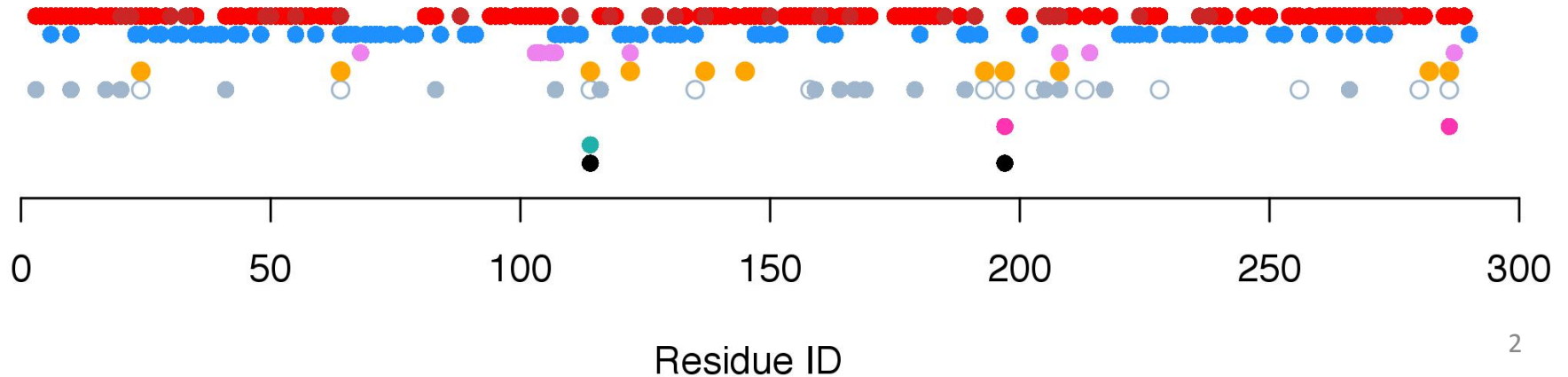
Gene Annotation	Gene Name
Cancer-related	NOTCH2; PDE4DIP; TPR; CRTC3; CDH11; MLLT6; ASXL1; HMGA1; KDM6A
DNA repair	RECQL; RAD51; PPM1D; XRCC1; AP1B1; FANCI; PTPRH; RBBP7; SLX4; POLR2A; DCLRE1C; ANKLE1
Cancer & DNA repair	ATM; PMS2; ERCC5
Actionable Gene	ATM; KDM6A; INSR; FOXP4

- **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

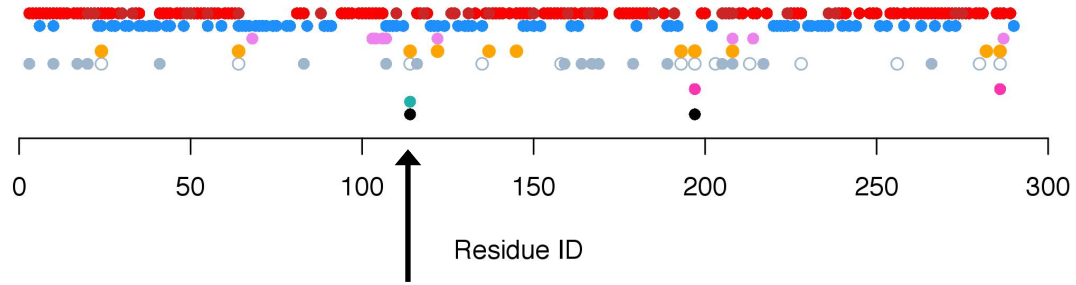


- Predicted allosteric (surface | interior)
- Buried residues
- Protein-protein interaction site
- Post-translational modifications
- ▲ ● HGMD (prem. stop | non-synon)
- ○ 1000 Genomes (rare | common)
- ▲ ● Snyder (prem. stop | non-synon)
- ▲ ● na12878 (prem. stop | non-synon)
- ▲ ● Subj. Z (prem. stop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

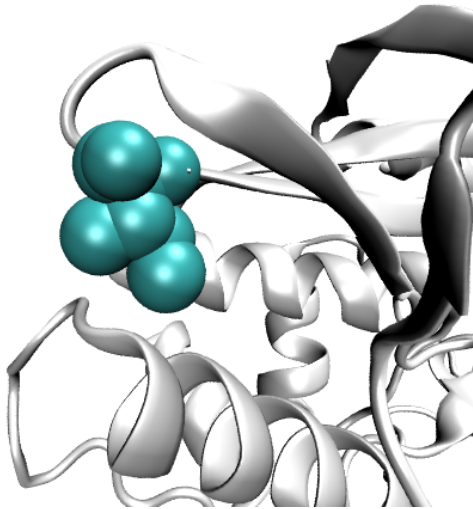
Arylamine N-acetyltransferase 2 (2PFR_A: gene = NAT2)



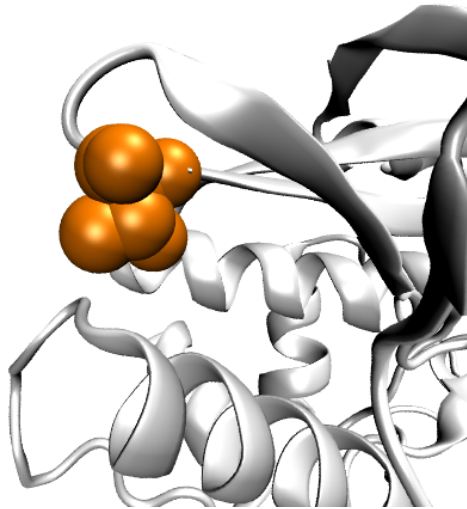
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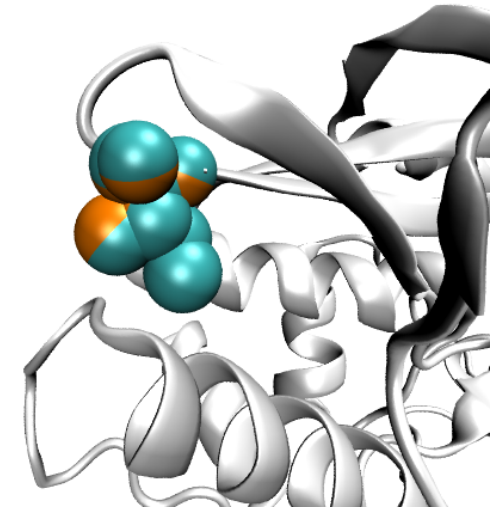
114: I->T



Wild-type

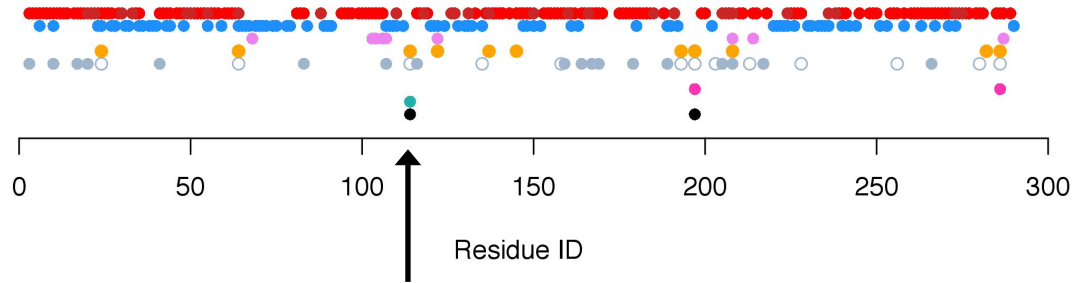


Mutated

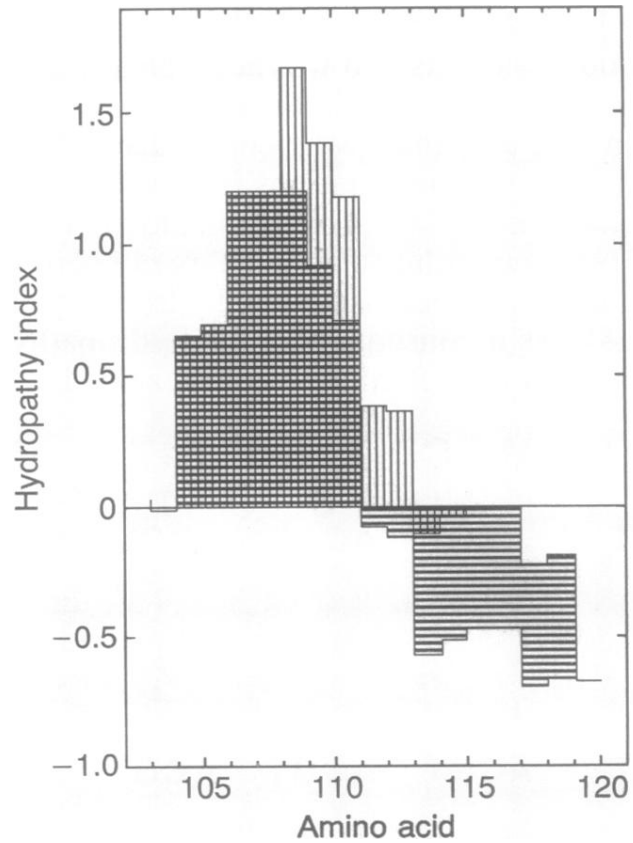


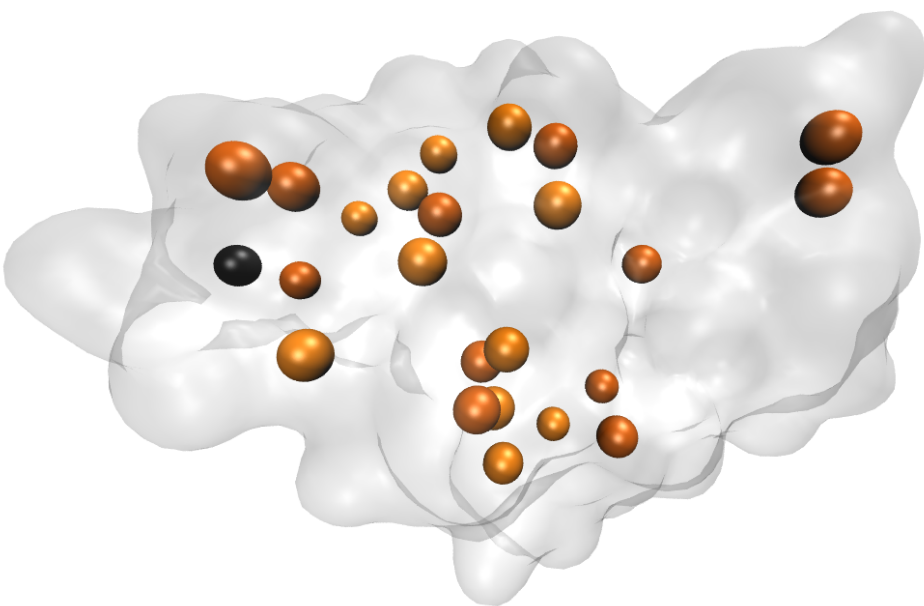
(superimposed)

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



114: I->T





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Tpr Domain of Aip (4APO_A: gene = AIP)

