

## 3D-CNN directly associates cognitive SNPs with brain structure



PRESENTER:  
**Leo Brueggeman**

### BACKGROUND

Genome-wide association studies of brain structure require extracting quantitative features from brain MRIs. This is technically challenging, limits the search space, and leads to high multiple testing burdens.

We show that **identifying SNPs associating with brain structure differences can be done directly using 3D Convolutional Neural Networks (3D-CNN)**, by predicting an individuals genotype from their raw brain MRI.

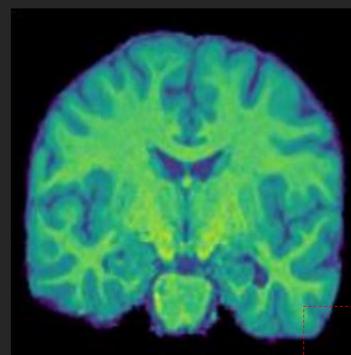
### METHODS

1. Collected **33K brain MRIs from UK Biobank** British ethnicity participants and their associated genotypes
2. SNPs previously reported to affect cognitive performance (CP), educational attainment (EA), and psychiatric disease (CD) were used
3. Sample was split into train, validation, and test sets
4. 3D-CNN models were trained to predict one SNPs genotype from brain MRIs
5. Test set values are corrected for scanner and participant covariates

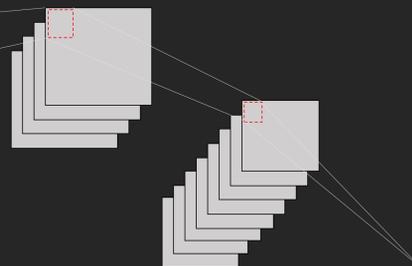
### RESULTS

- 3 of 220 SNPs exceed validation p-value of 0.0015
- 1 of these SNPs had a highly significant association after correction for covariates in test set (rs13107325)

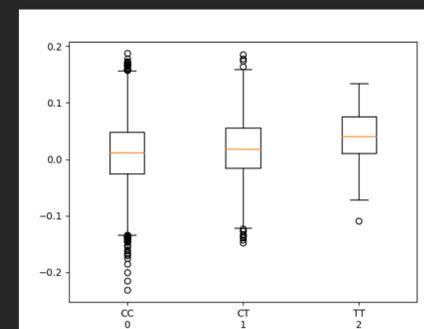
# Deep learning can predict your genotype directly from brain structure



1) RAW MRI



2) 3D-CNN MODEL



3) GENOTYPE PREDICTION



Take a picture to see the poster online

### RESULTS

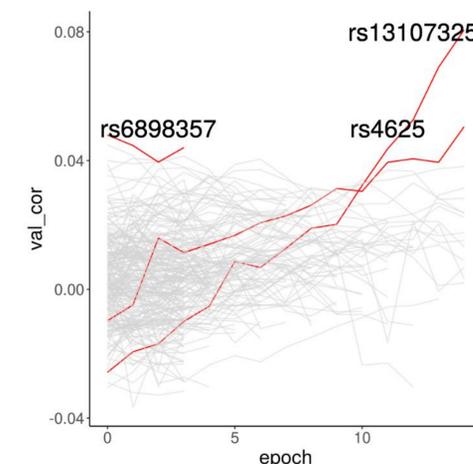


Fig 1. Validation Pearson Correlation of all SNPs tested over epochs

STUDY	Rsid	Gene	Epoch	Val Cor	Val p	Test Cor	Test p
CP	rs13107325	SLC39A8	15	0.08	1.1e-8	0.085	2.9e-17
CP + EA	rs4625	DAG1	15	0.05	3.5e-4	-0.01	0.20
CD	rs6898357	TENM2	4	0.04	1.5e-3	0.008	0.27

Table 1. Validation and test set performance of 3 SNPs found to have high significance ( $p < 1.5e-3$ ) in the validation set

### FUTURE DIRECTIONS

- Extended testing of 4000 SNPs total
- Extend to include SNPs from other traits, such as brain region volumes and individual psychiatric diseases
- Model architecture and hyperparameter optimization
- Visualization of brain regions used for genotype prediction

Tanner Koomar, Jacob Michaelson

