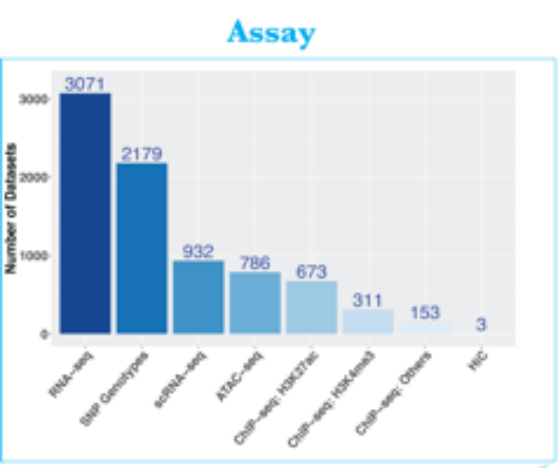

Assay
Number of Datasets
3000
2000
1000
0
3071
2179
932
786
673
311
153
3
RNA−seq
SNP Genotypes
scRNA−seq
ATAC−seq
ChIP−seq: H3K27ac
ChIP−seq: H3K4me3
ChIP−seq: Others
HiC

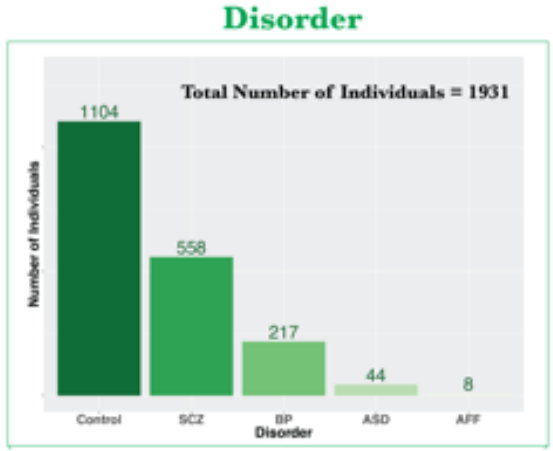
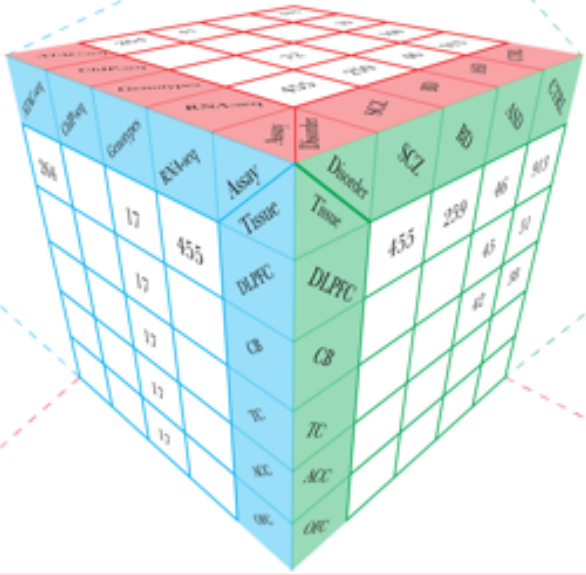
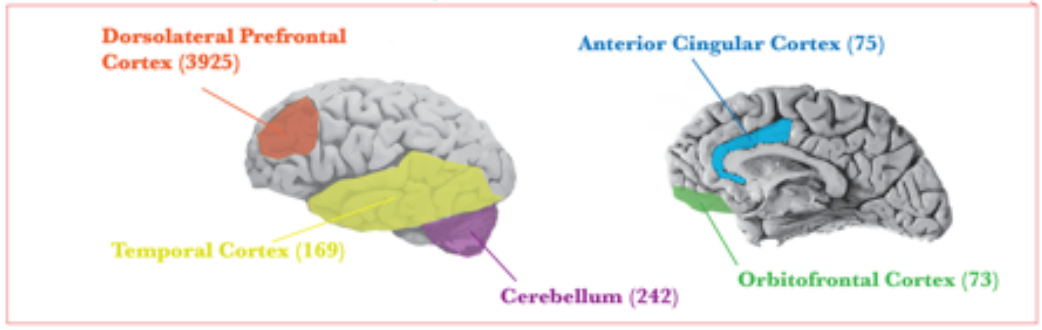
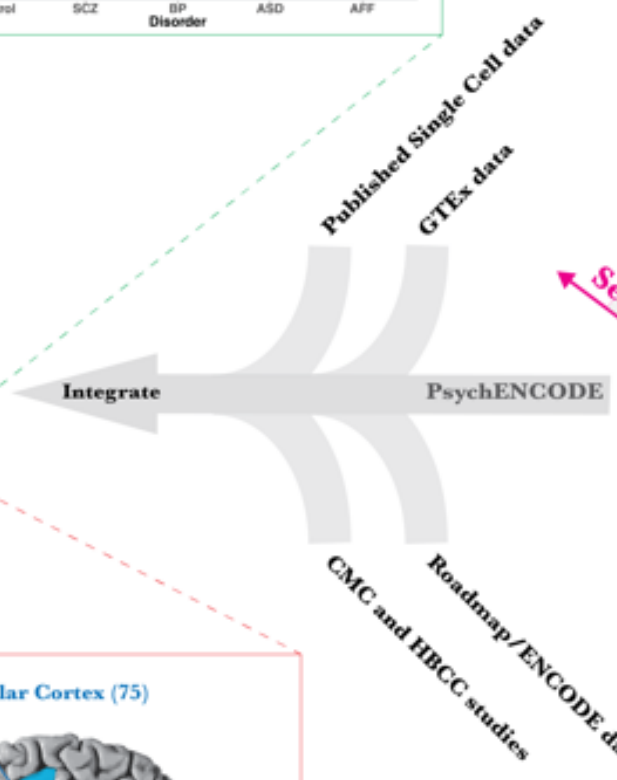
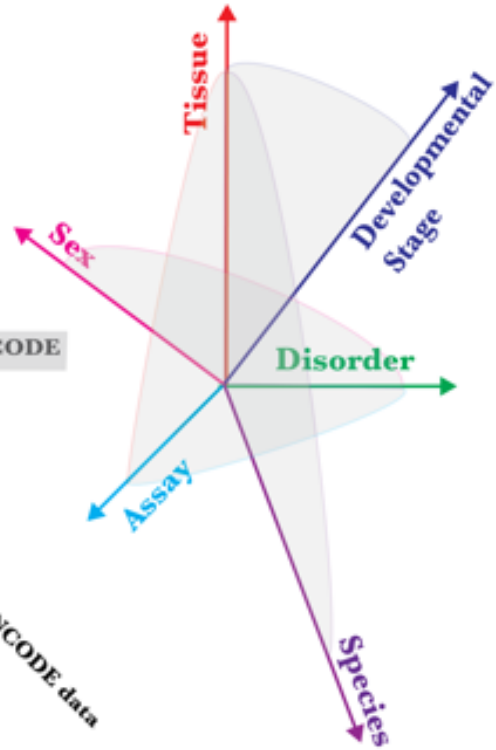

Disorder
Total Number of Individuals = 1931
Number of Individuals
1104
558
217
44
8
Control
SCZ
BP
ASD
AFF
Disorder

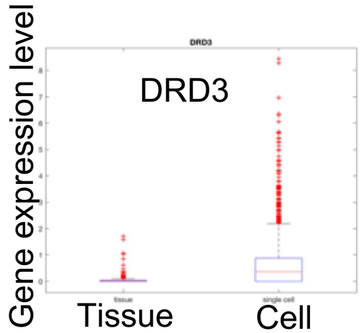
Figure 1




Dorsolateral Prefrontal Cortex (3925)
Anterior Cingular Cortex (75)
Temporal Cortex (169)
Cerebellum (242)
Orbitofrontal Cortex (73)
Tissue


Published Single Cell data
GTEx data
Integrate
PsychENCODE
CMC and HBCC studies
Roadmap/ENCODE data

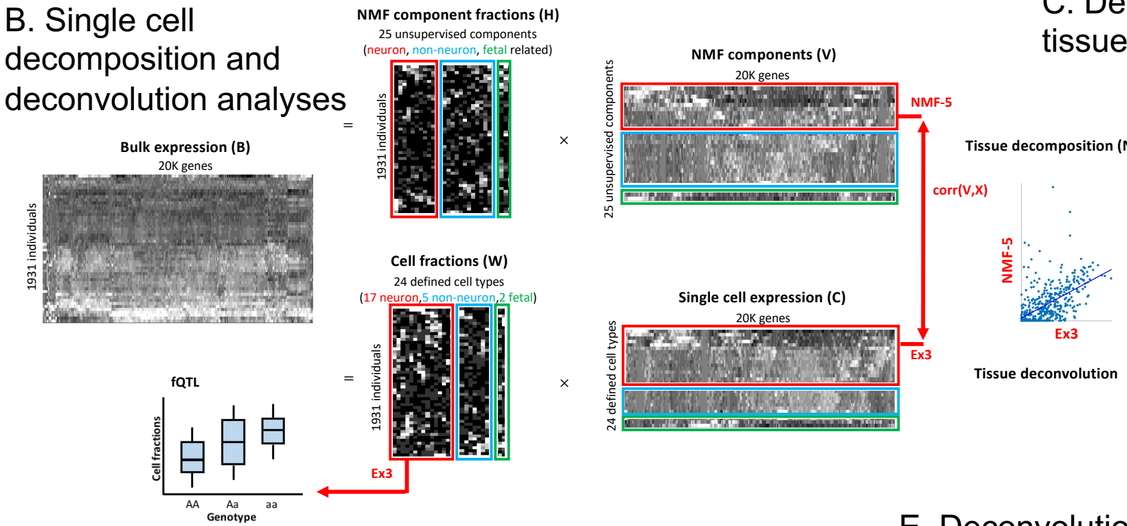

Tissue
Developmental Stage
Sex
Disorder
Assay
Species



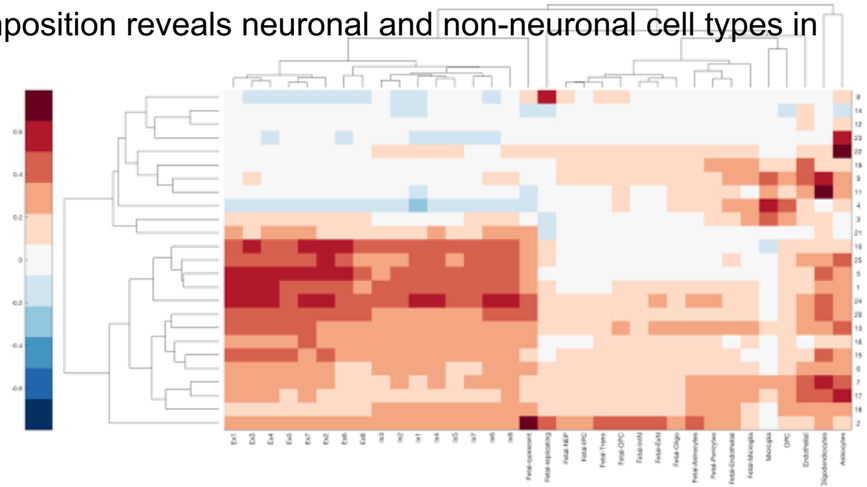
A. Neuronal gene expression variations of tissues vs. cells

Figure 2 Bulk and Single Cell transcriptome

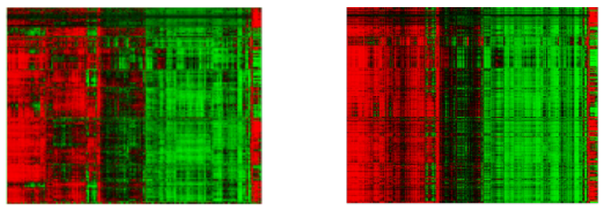
B. Single cell decomposition and deconvolution analyses



C. Decomposition reveals neuronal and non-neuronal cell types in tissues



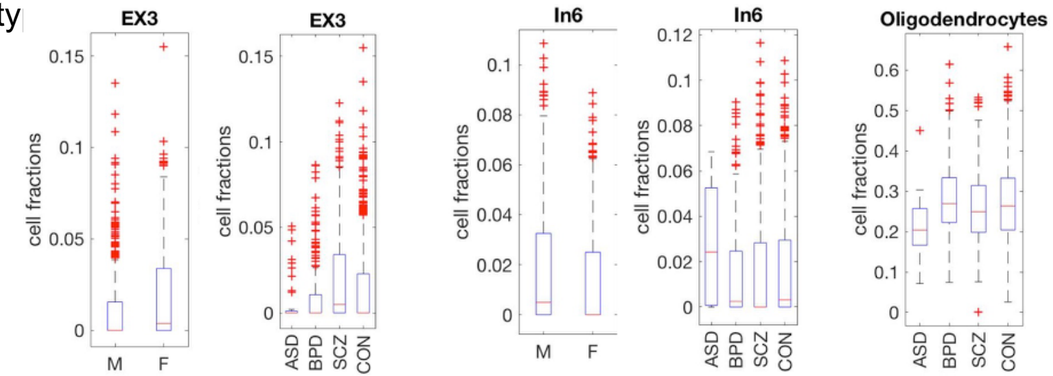
Builk expression (B)



Fitted expression by deconvolution (W*C)

D. Single cell gene expression explains >80% tissue expression covariation

E. Deconvolution reveals neuronal and non-neuronal cell populations of phenoty



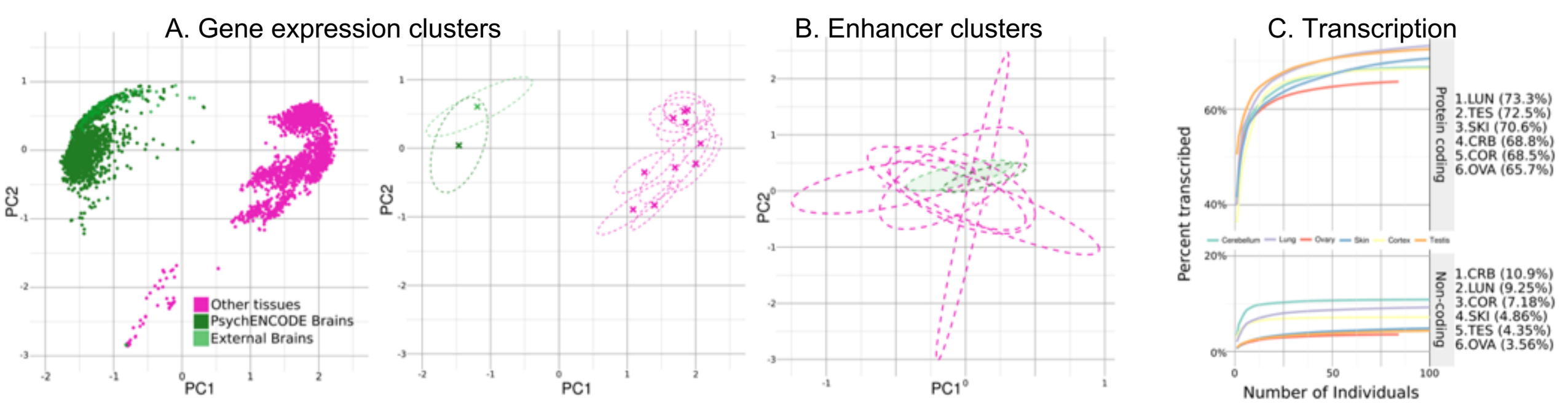
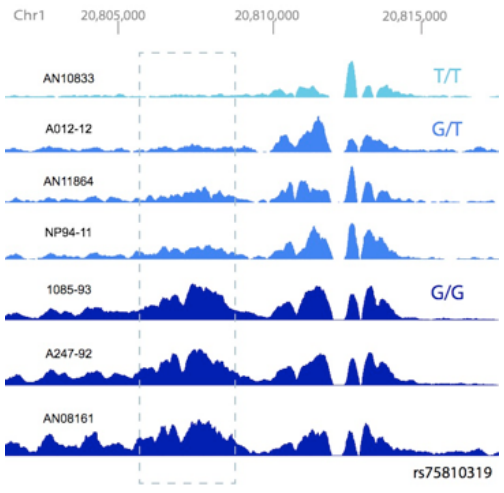
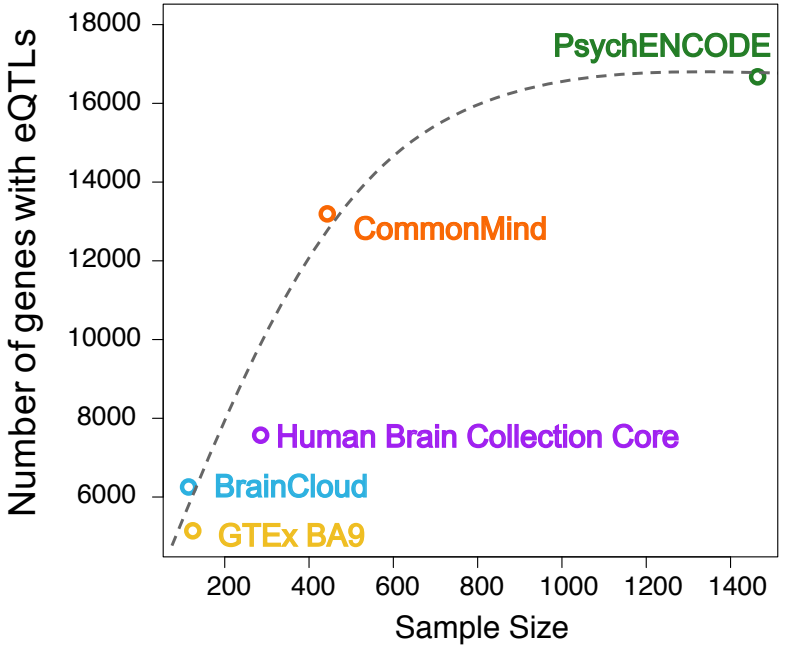


Figure 3. Comparative analysis

A. QTLs



B. # of eGenes



C. Stats about QTLs

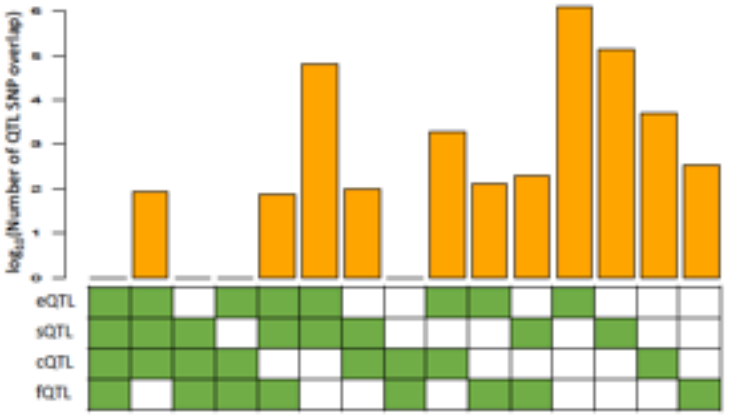
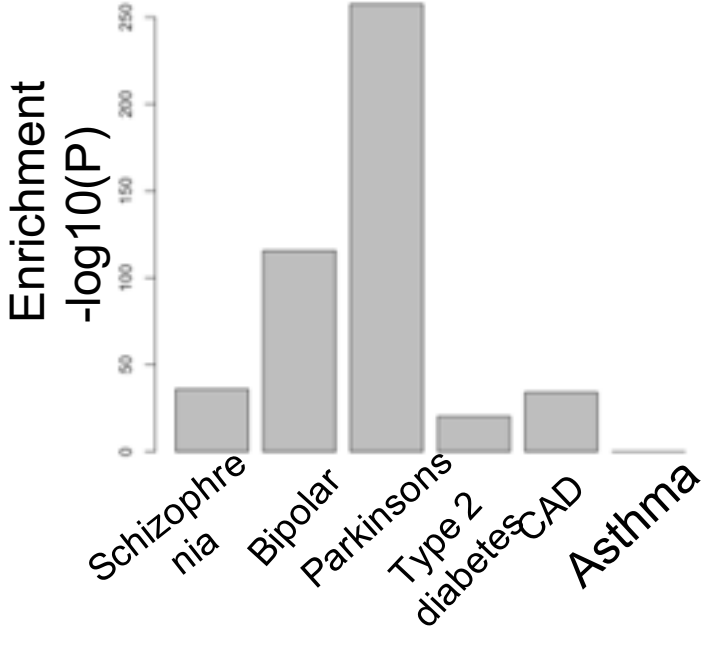
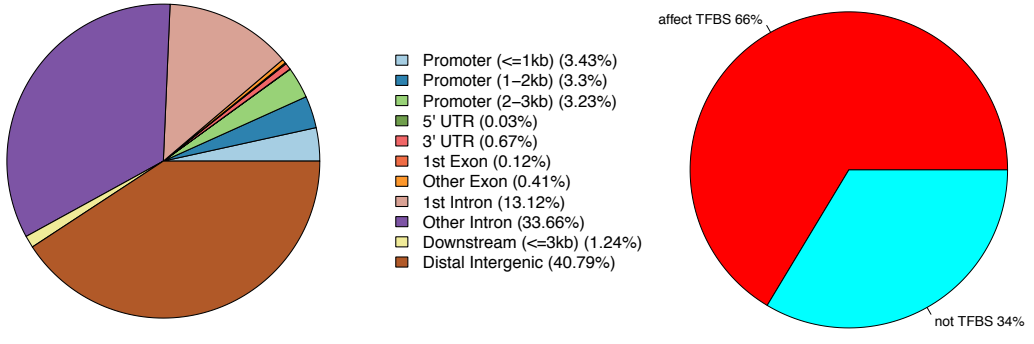


Figure 4
QTL analysis

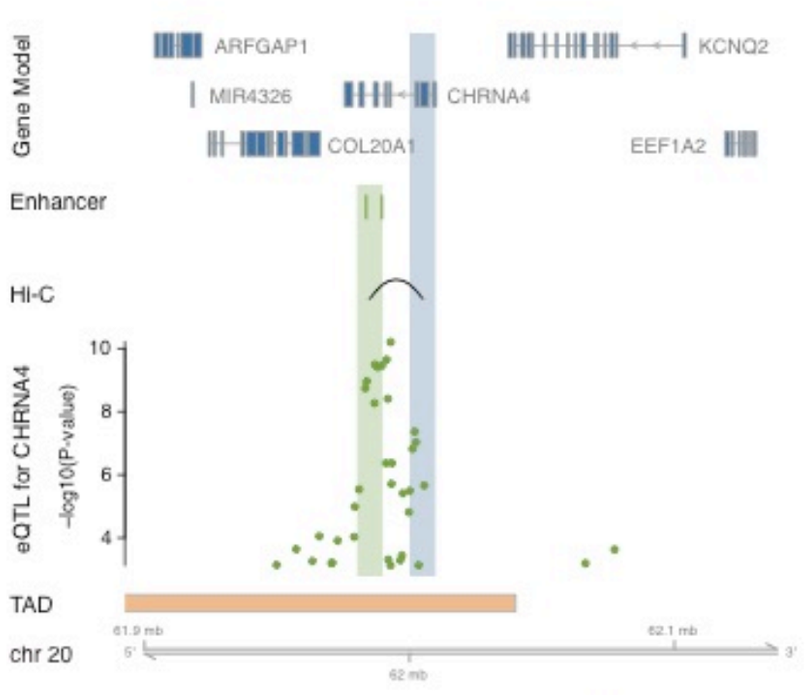
D. Comparison with GWAS



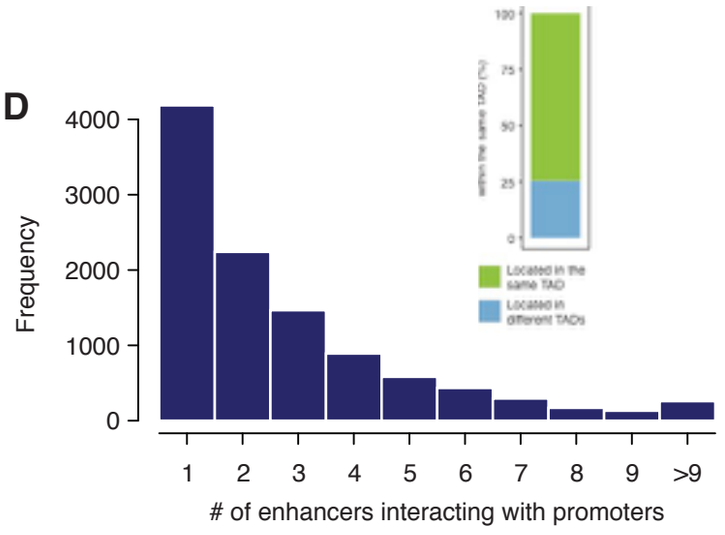
E. Annotations of eQTLs



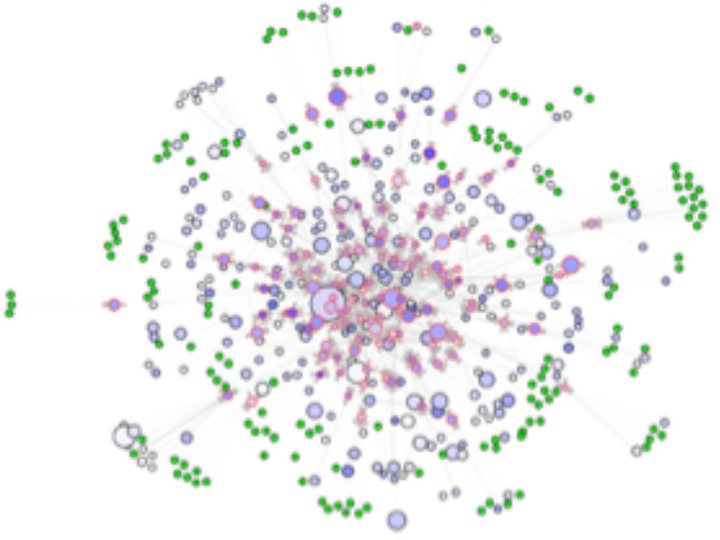
A. Hi-C



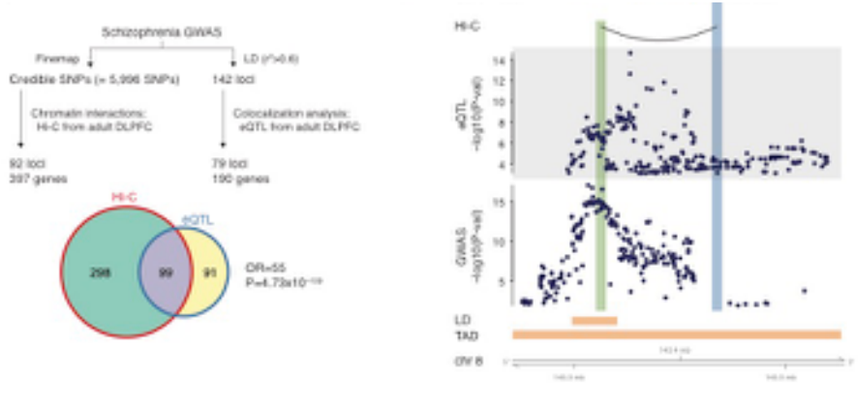
B. enhancer-target linkage



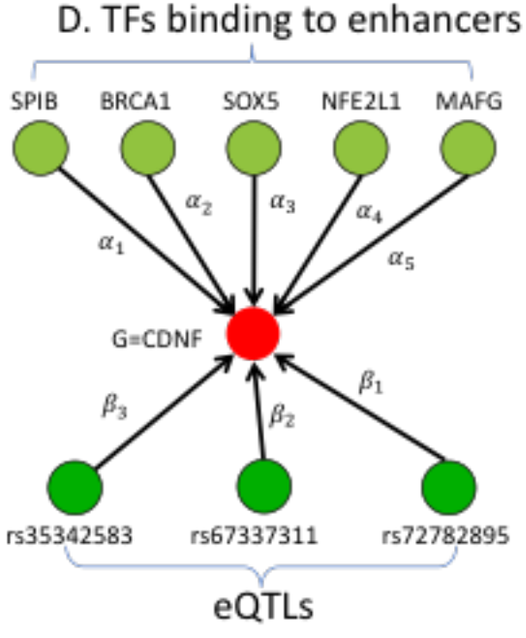
E. Gene regulatory network



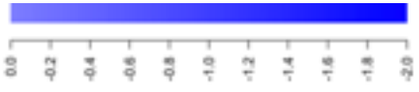
C. HiC vs. eQTL



D. TFs binding to enhancers



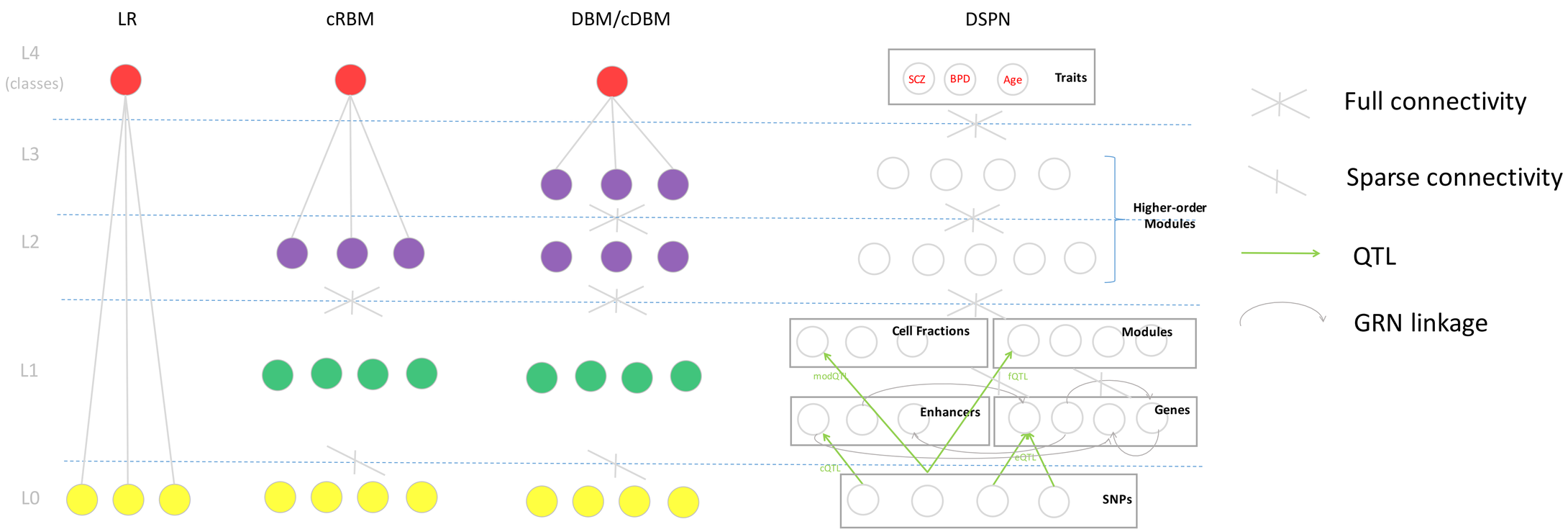
Gene expression fold change for phenotype



- Brain gene (GTEx)
- eQTL and cQTL
- genes with highly/lowly phenotype-associated eQTLs

Figure 5 network

(a) schematic



(b) Full model performance:
% accuracy on balanced sample

	SCZ	BIP	ASD	Gender	Ethnicity	Age (>51)
Gen (LR)	54.6%	56.7%	50%	50%	99.0%	61.9%
Tr (LR)	63.0%	63.3%	51.7%	69.7%	86.0%	81.2%
Tr (DSPN)	73.6%	76.7%	73.3%	72.1%	87.7%	86.2%

Gen = Genome-based; Tr = Transcriptome-based; LR = Logistic Regression; DPSN = Deep Structured Phenotype Network

(c) Transcriptome and epigenome imputation; % accuracy (binarized)

Expression

# genes	All	1000	500	250	100	50	10
Test accuracy	52.7	55.0	56.9	59.2	63.1	66.3	72.9

Enhancers

# genes	All	1000	500	250	100	50	10
Test accuracy							

Prediction with imputation

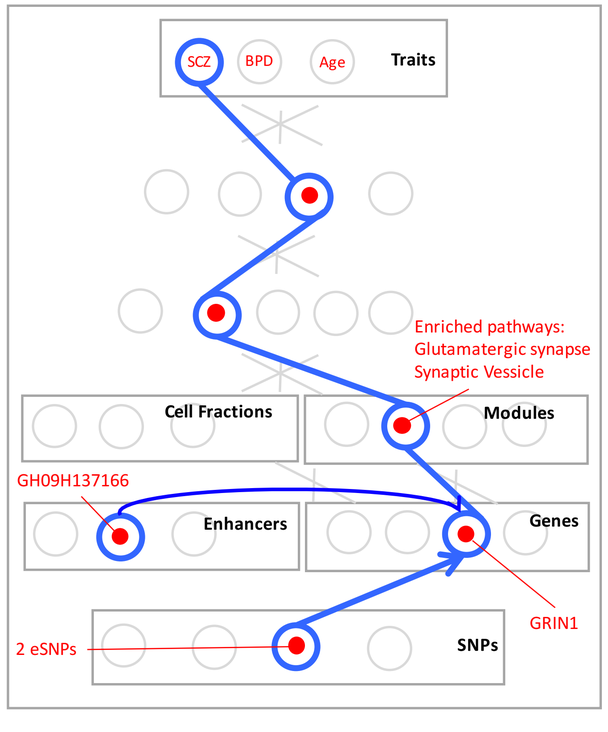
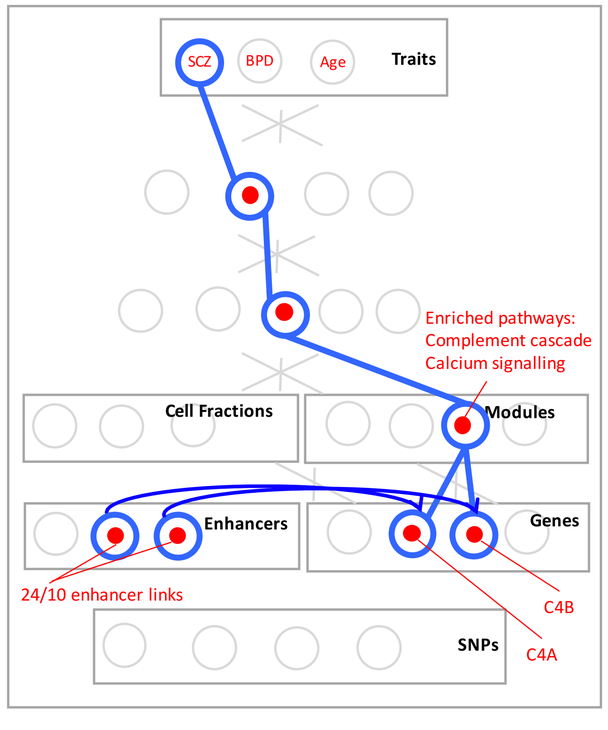
SCZ	BIP	ASD
57.9%	66.7%	65%

(d) Imputation-based vs Full model:
% variance explained on liability scale

	Gen (LR)	Imputed (eQTL only)	Imputed (fQTL only)	Imputed (cQTL only)	Imputed (all)	DSPN (full model)
SCZ	0.5%	1.3%	4.5%	-	4.5%	32.8%
BIP	3.4%	16.3%	8.7%	-	22.6%	53.8%
ASD	0%	0.4%	0%	7.4%	7.4%	21.4%

(e) Functional analysis

GRIN1



C4A/C4B