

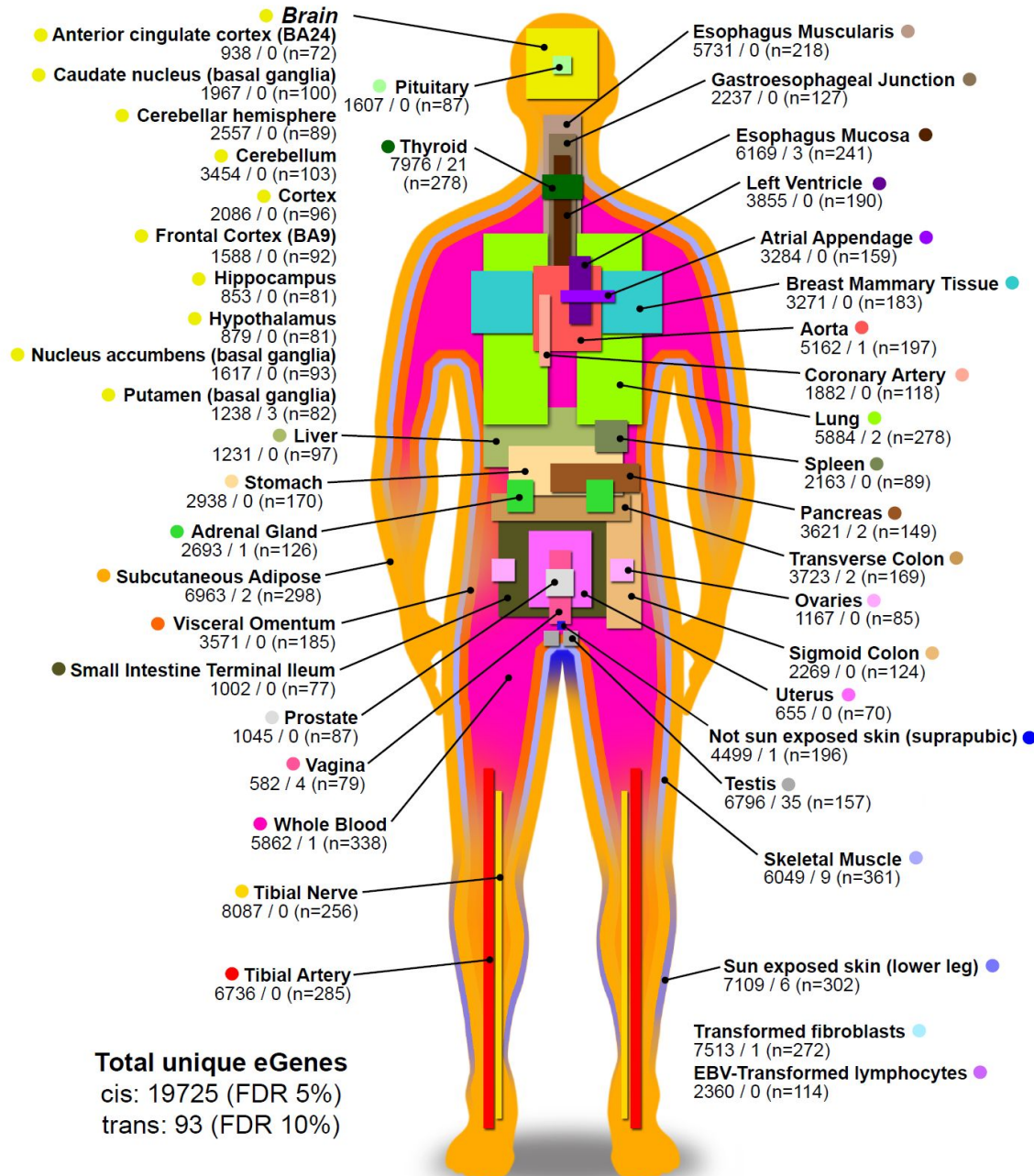


GTEx project updates: intersecting epigenomes and eqtls

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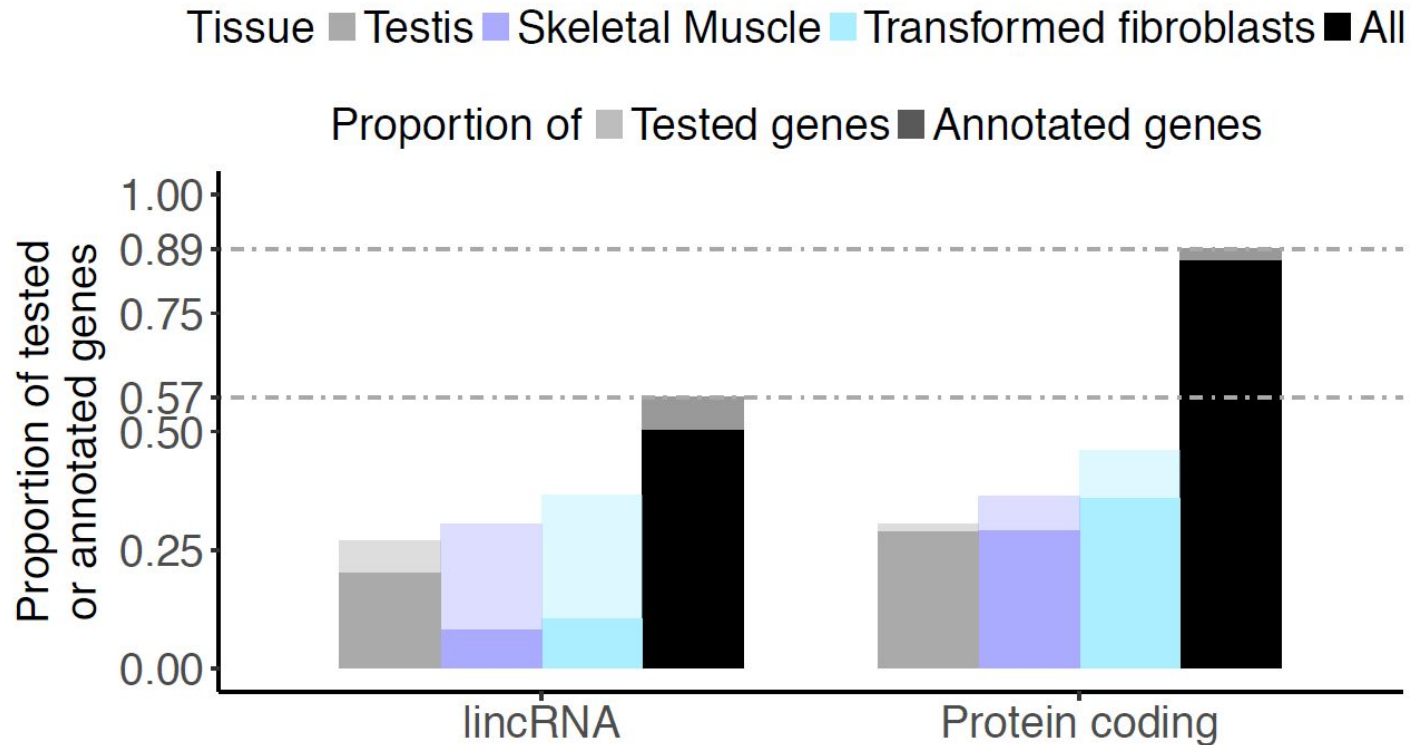
Reference map of eQTLs across multiple human tissues

version 6 (in press)
 7,051 samples from 449 donors

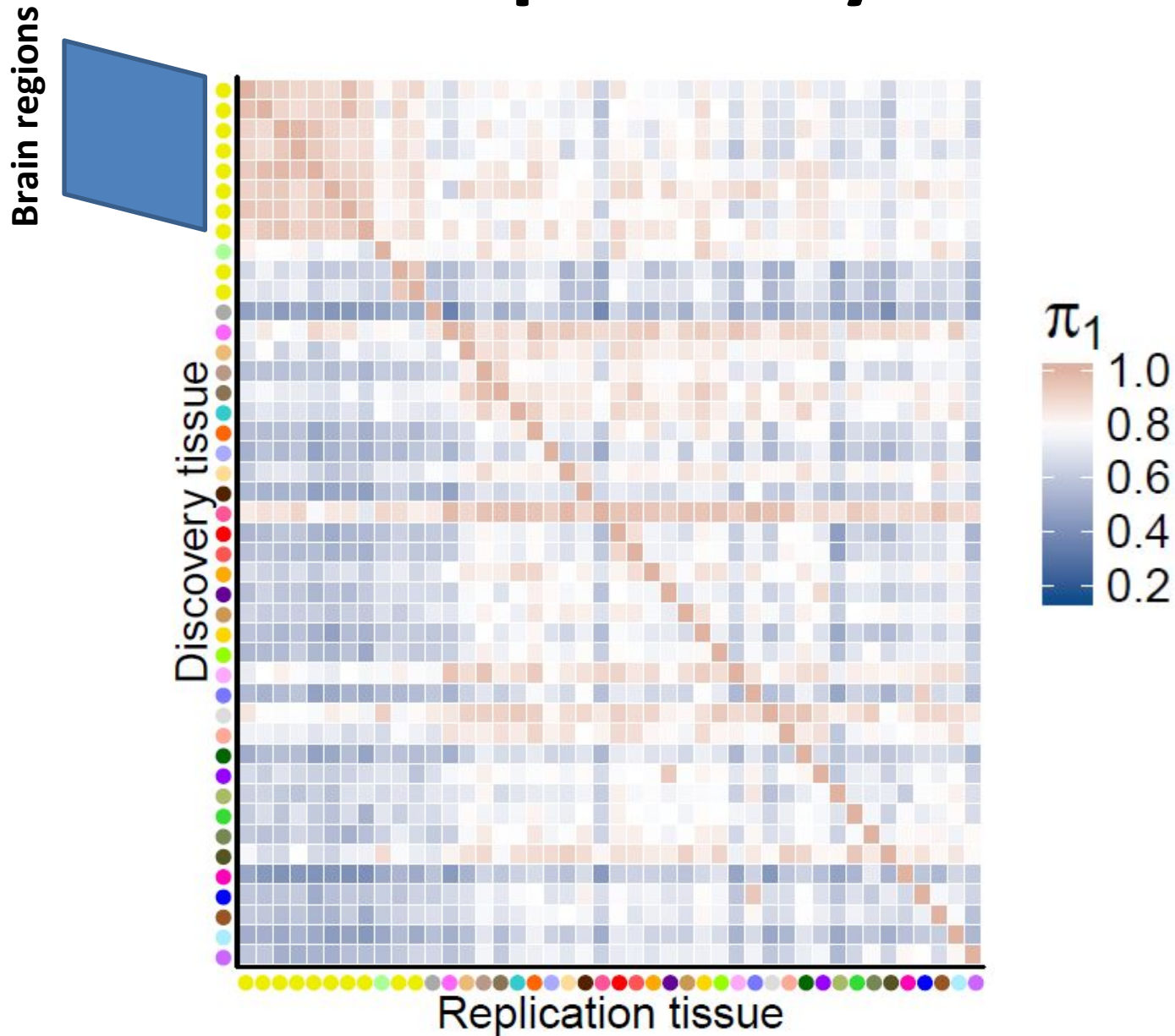
version 7
 11,688 samples from 635 donors

version 8 (in QC)
 17,382 samples from 838 donors

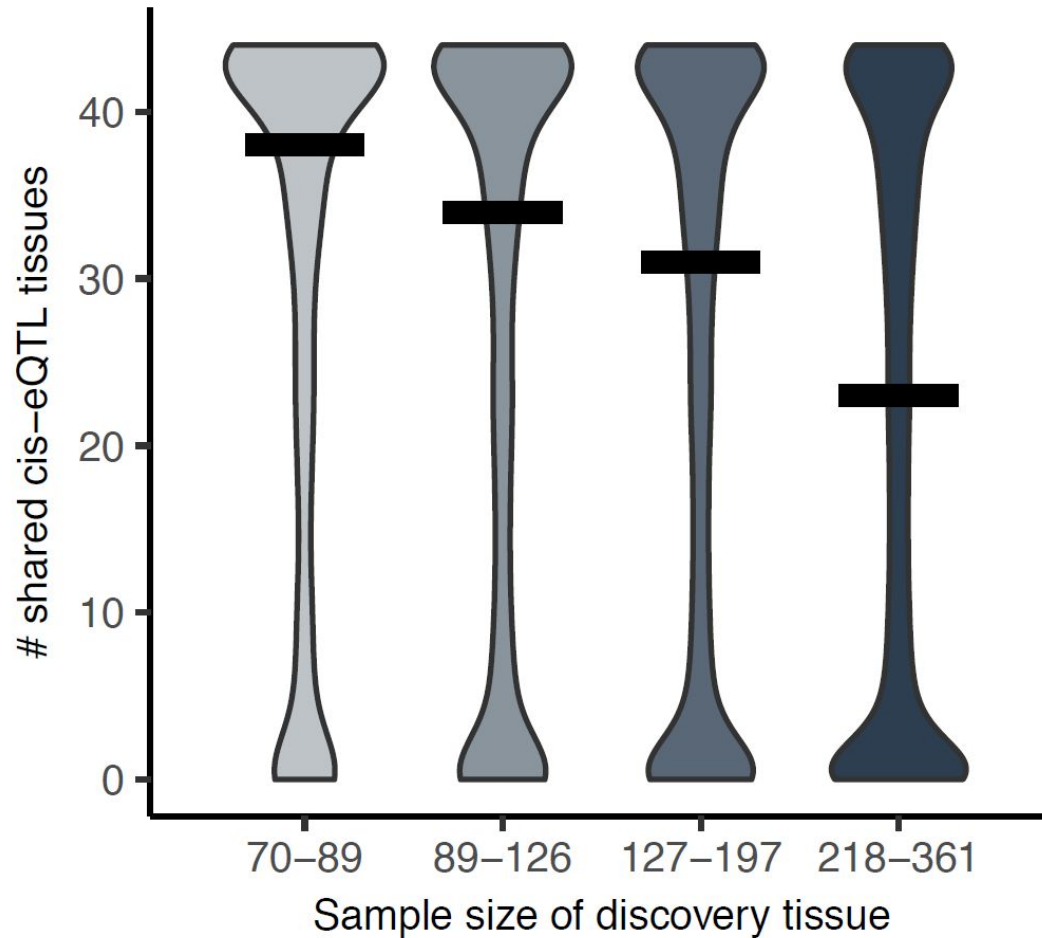
Nearly all known human genes have expression influenced by genetics



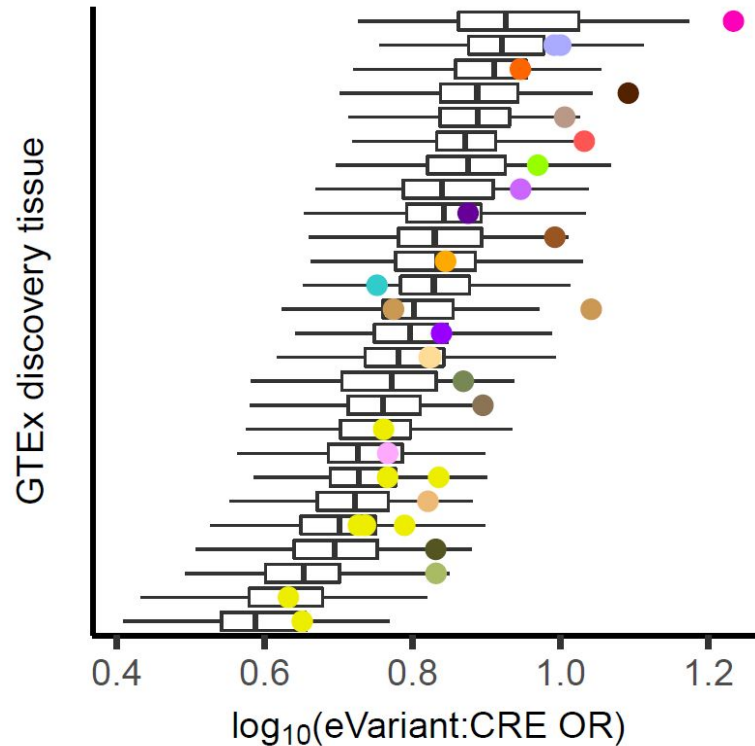
Tissue-specificity of eQTLs



Bimodal pattern of eQTL sharing

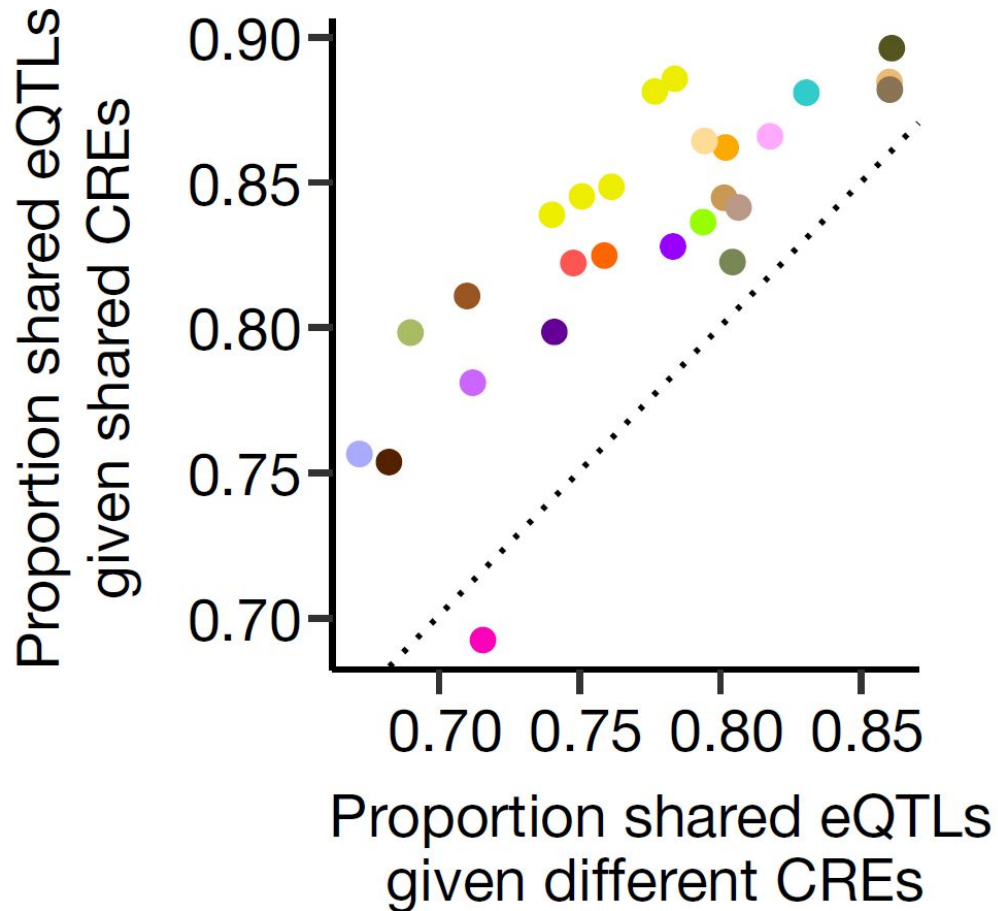


Tissue-eQTLs enriched in matched tissue epigenomes

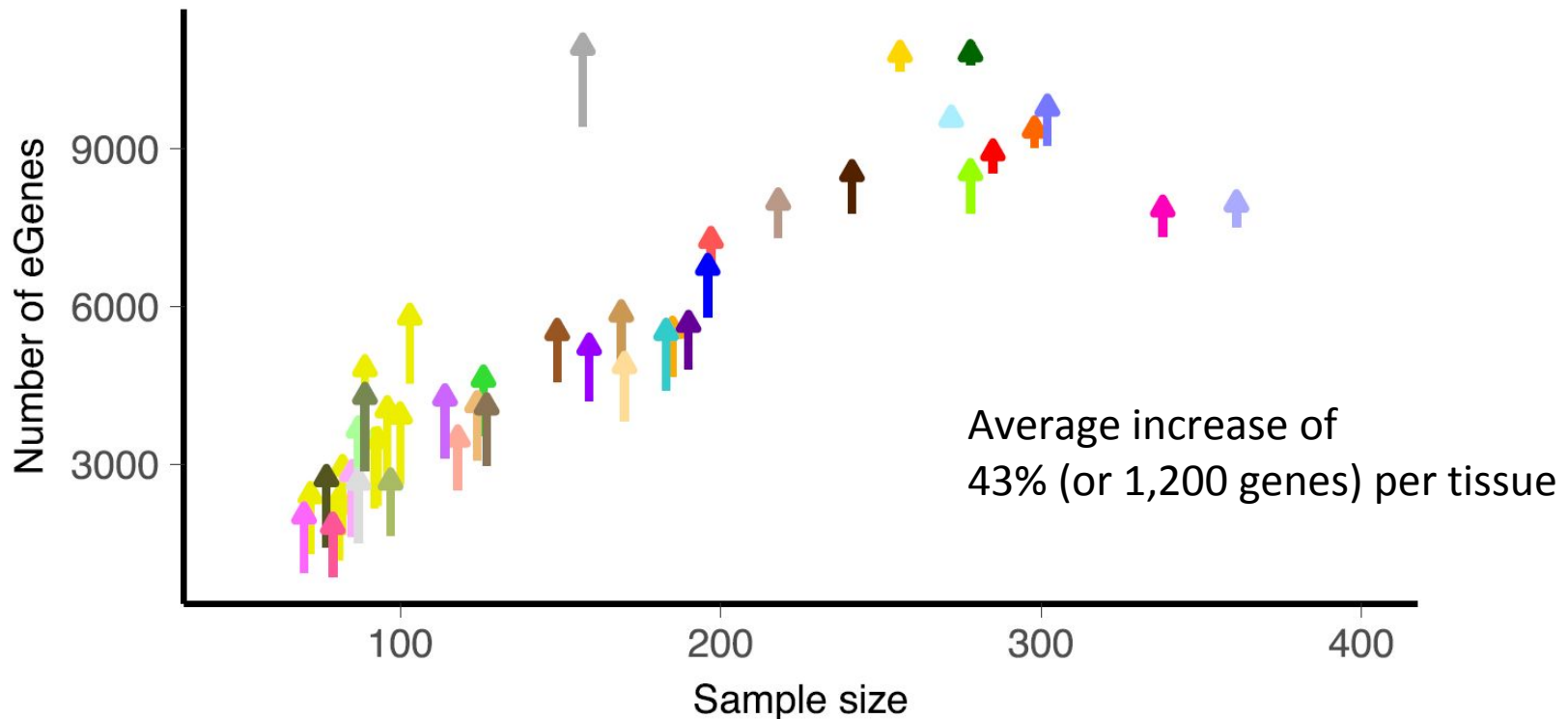


Enrichment (x-axis) of eVariants in cis-regulatory elements (CREs) across 128 Roadmap Epigenomics project cell types, for each GTEx discovery tissue (y-axis). Enrichment estimated by comparing to random MAF and distance-matched variants. Stronger enrichment was observed in matched tissues (coloured dots) than in unmatched tissues (box plots).

CRE sharing reflects eQTL sharing



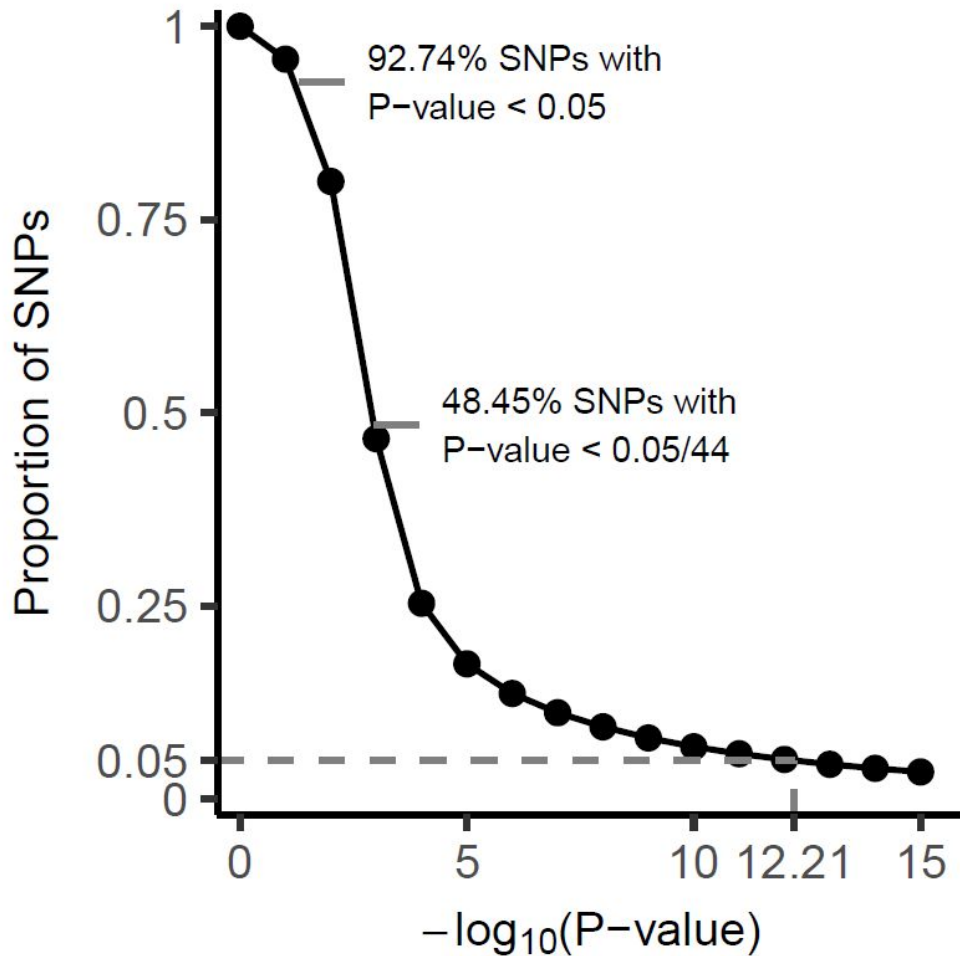
Combining functional annotation with eQTL discovery



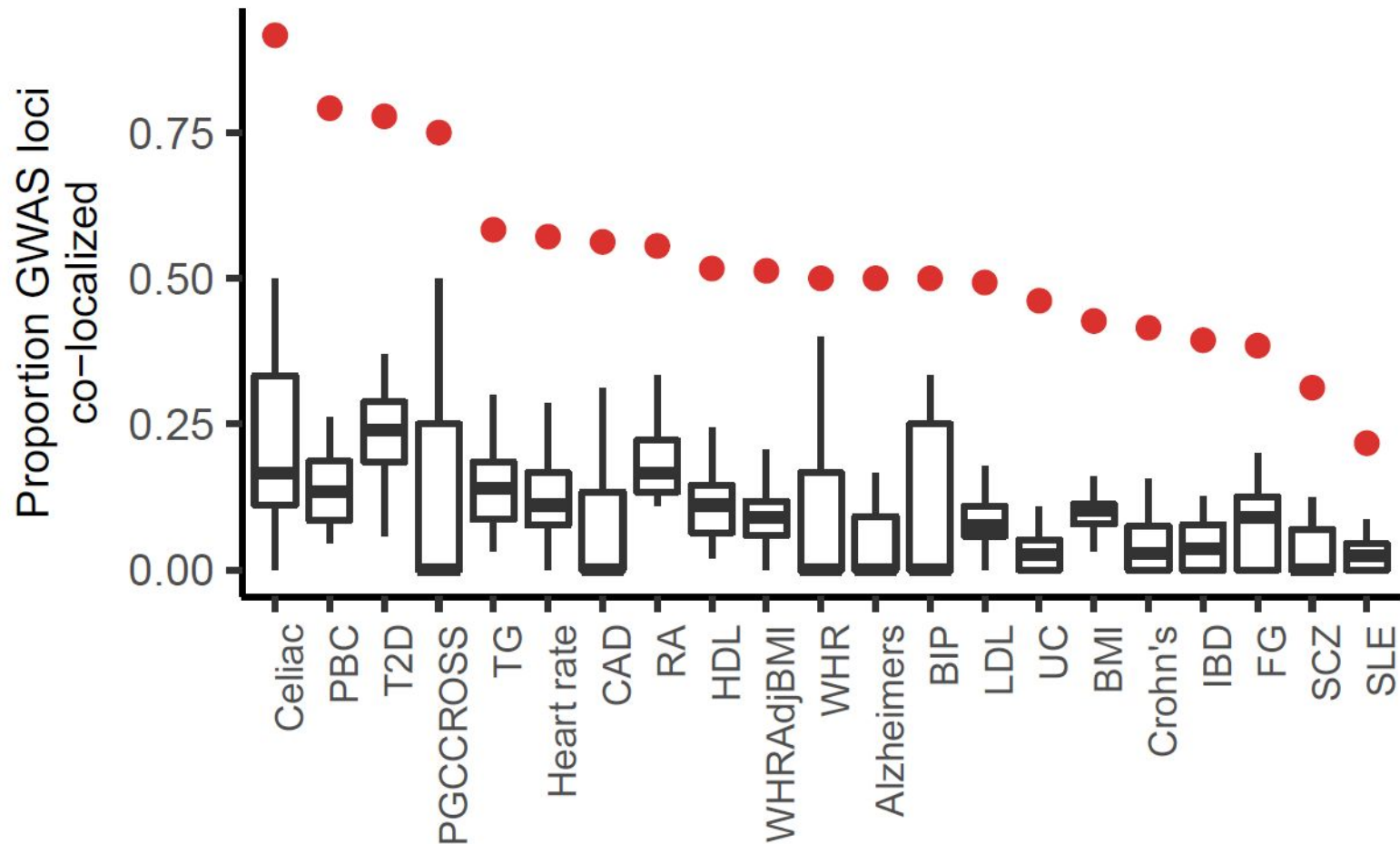
Wen, X., Lee, Y., Luca, F. & Pique-Regi, R. Efficient integrative multi-SNP association analysis via deterministic approximation of posteriors. *Am. J. Hum. Genet.* **98**, 1114–1129 (2016).

Implications of GTEx for interpreting GWAS signals

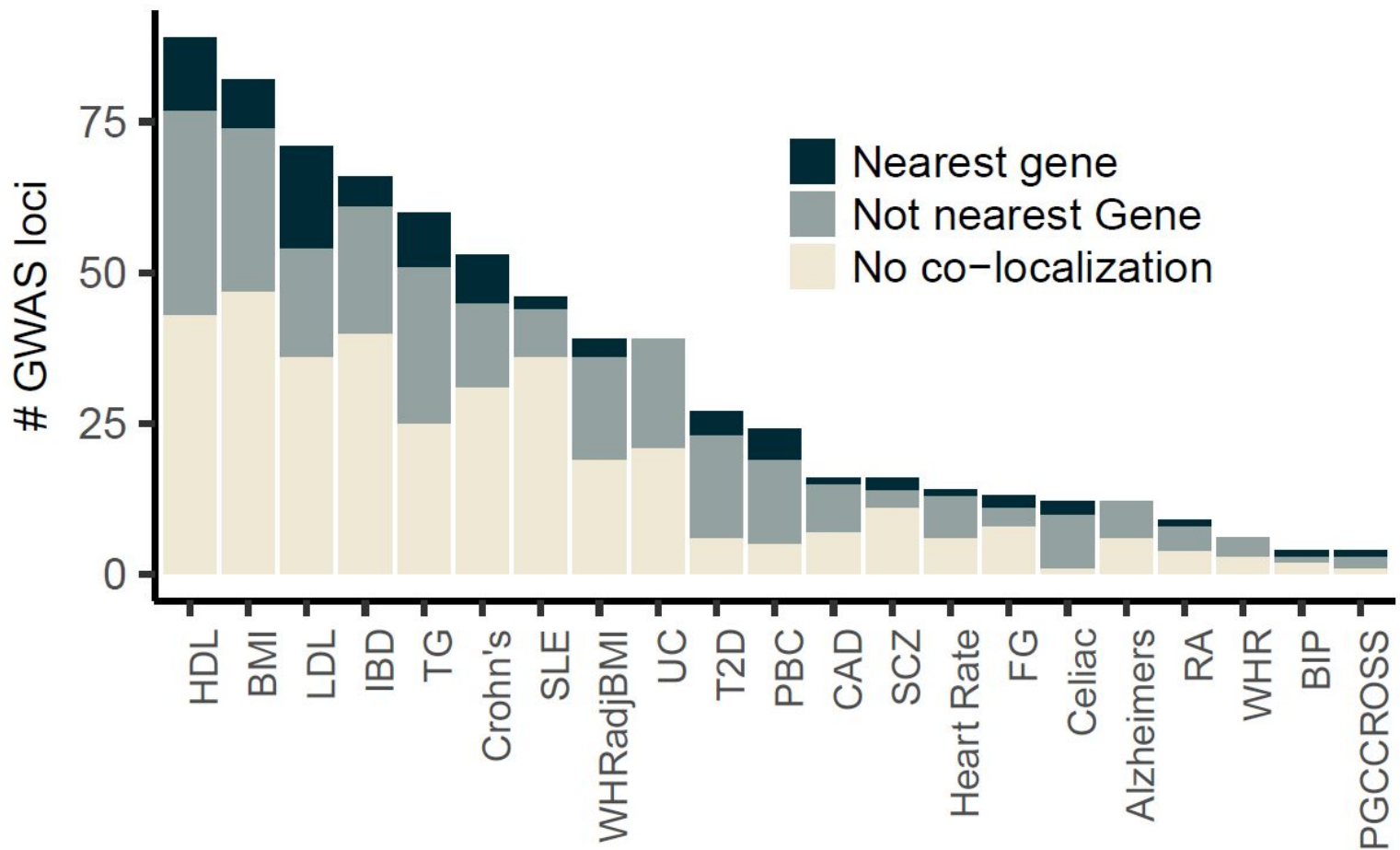
Lots of eQTL data means that seemingly significant associations are the norm



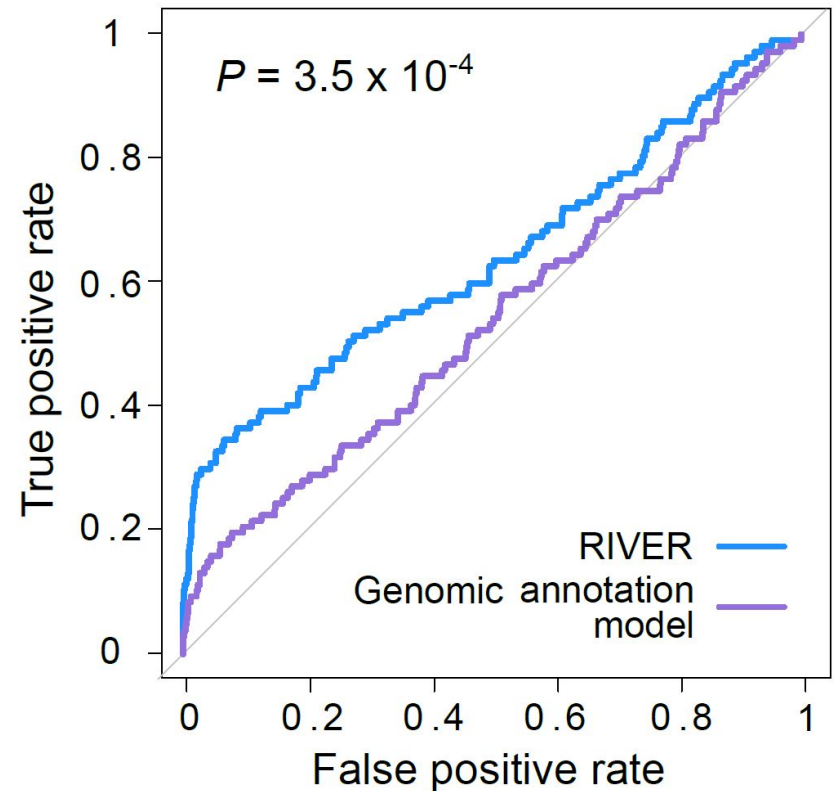
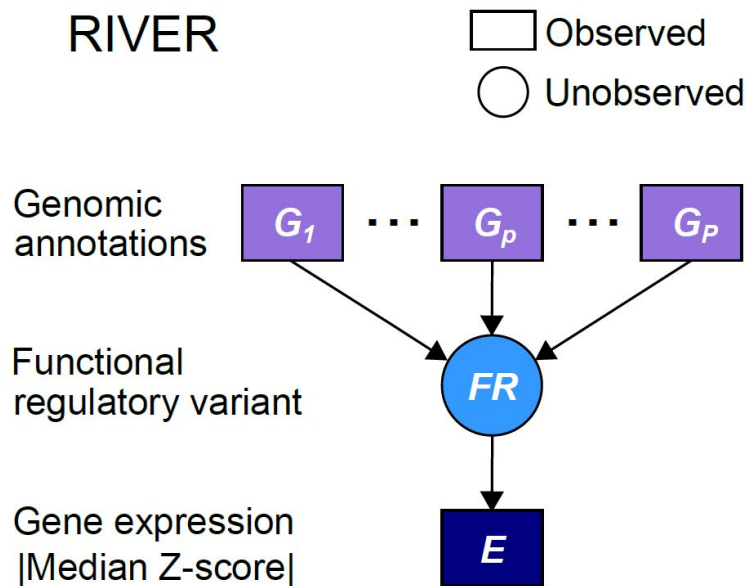
Co-localization of eQTLs and GWAS in GTEx



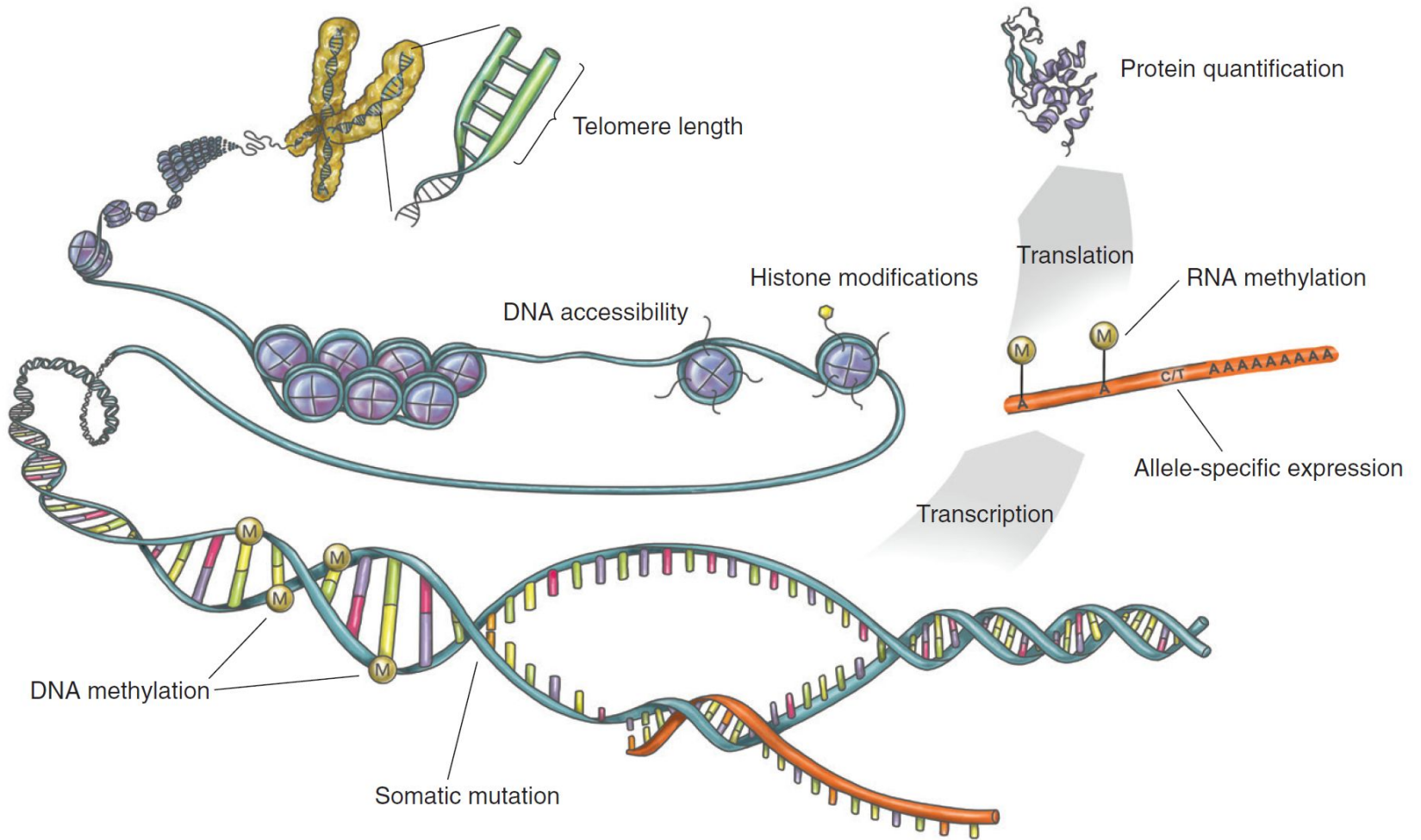
Co-localization of eQTLs and GWAS in GTEx



Interpreting personal variants using genetic and functional genomics data



eGTEx project



eGTEx project

Table 1 eGTEx study design

Molecular phenotype	Primary assay(s)	Targeted tissues (phase II)	Targeted sample number
DNA accessibility	DNase I hypersensitivity	Brain regions, heart, lung muscle, esophagus, breast, prostate, skin	~1,135
Histone modifications	ChIP-seq	Brain regions, heart, lung, muscle	~600
DNA methylation	WGBS and capture bisulfite sequencing	Brain regions, heart, lung, muscle, thyroid	~2,000
Allele-specific expression	mmPCR-seq	All tissues	~2,000
Post-transcriptional RNA modifications	m ⁶ A methylation capture sequencing	Brain regions, heart, lung, muscle	~300
Proteomic variation	MS, targeted arrays for transcription factors and cell signaling proteins	Brain, heart, lung, muscle, thyroid, colon, liver, prostate, pancreas, ovary, testis, breast	~1,000 (MS) ~2,500 (arrays)
Somatic variation	Deep exome sequencing, RNA-seq, SNP arrays, probe-based telomere length assay	~20–25 tissues	~800
Telomere length	Luminex-based assay for telomere-repeat abundance	~20 tissues	~5,000

Molecular assays, targeted tissues, and sample number for eGTEx.

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