

The GTEx Consortium atlas of genetic regulatory effects across human tissues

汇报组：2组

秦秋羽 王梓聿 张晨 李奕昕 蒲明怡 王聪

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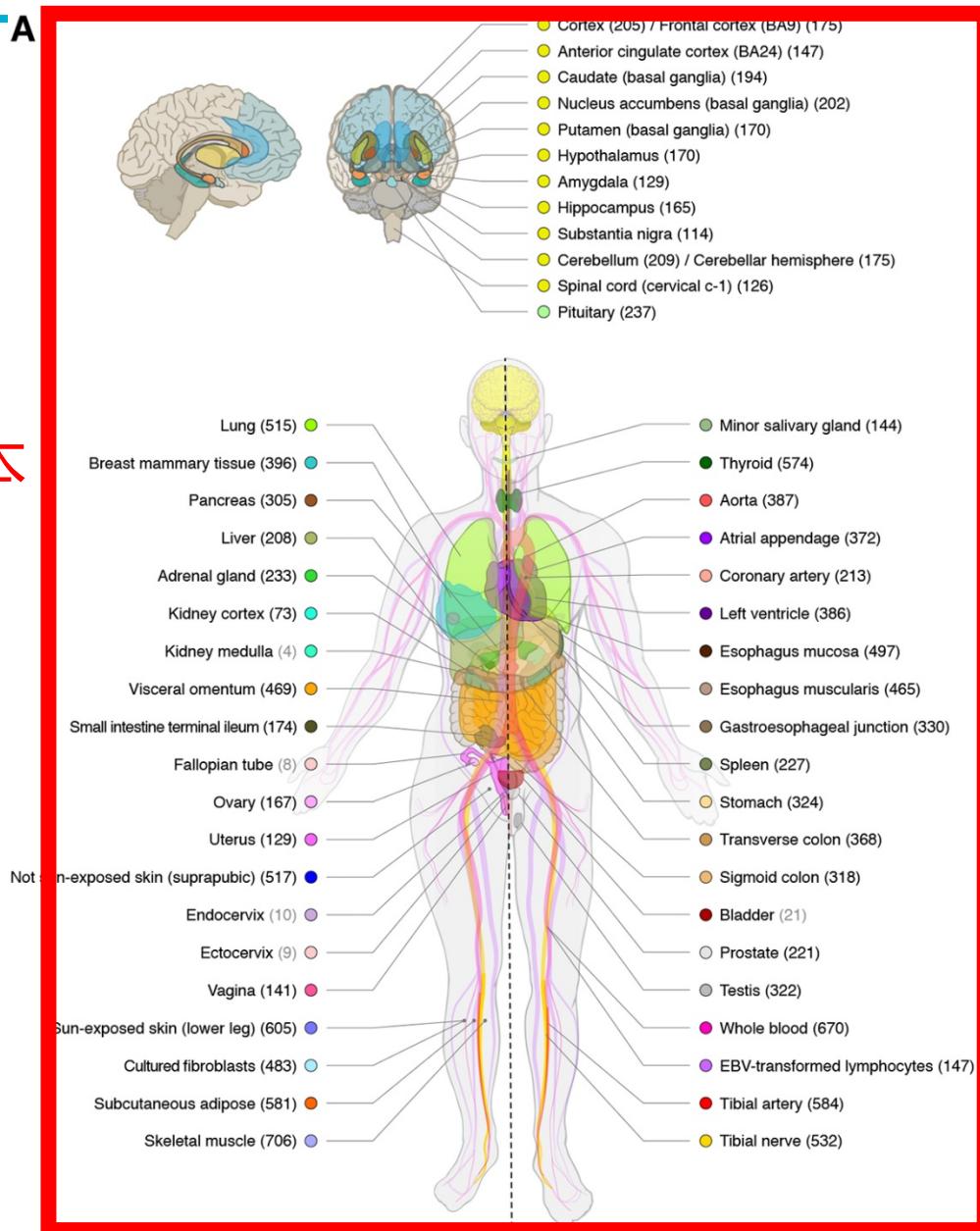
The Genotype-Tissue Expression (GTEx) project:

- Launched in 2010
- Including 838 post-mortem donors, 49 tissues, 15,201 RNA-sequencing samples
- Links genetic effects on the transcriptome to disease associations

Fig1. Sample and data types in the GTEx v8 study

质控后样本
组织类型

A



B

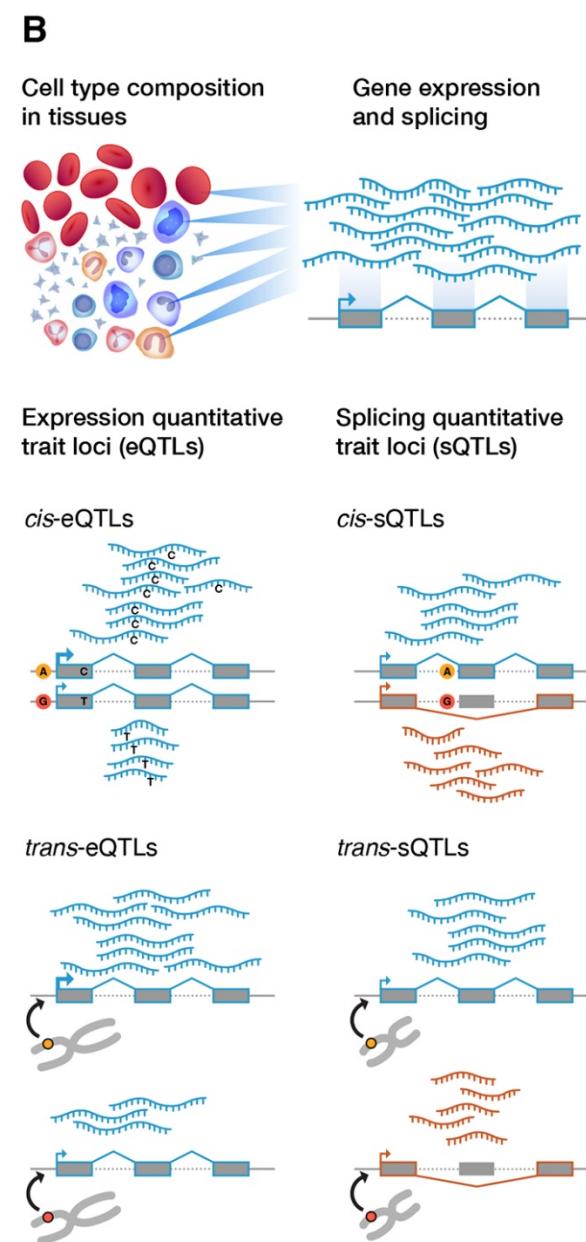


Fig1. Sample and data types in the GTEx v8 study

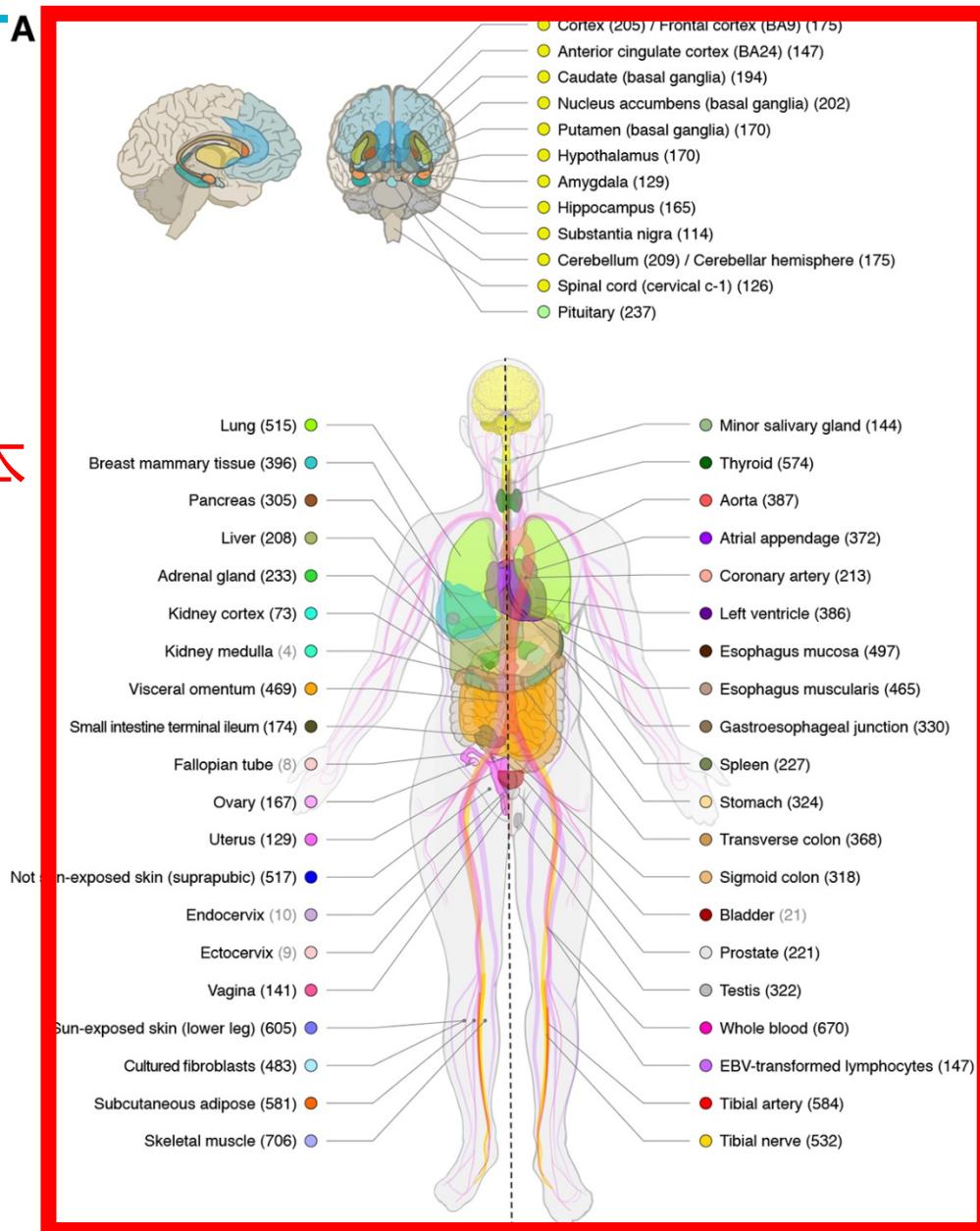
Quantitative trait locus (QTL): 数量性状基因座

- eQTLs: 能够控制数量性状基因表达水平高低的基因组区域。
- sQTLs: 能够控制数量性状基因可变剪接水平高低的基因组区域。

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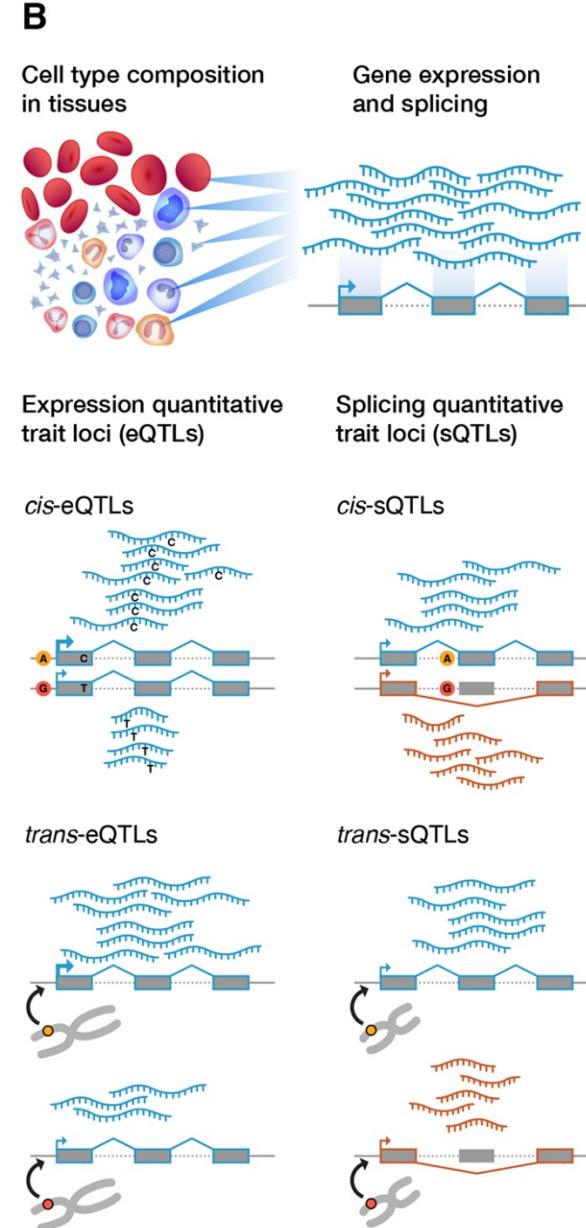


Fig2. QTL discovery

具有eQTL或sQTL的基因
被称为eGenes和sGenes

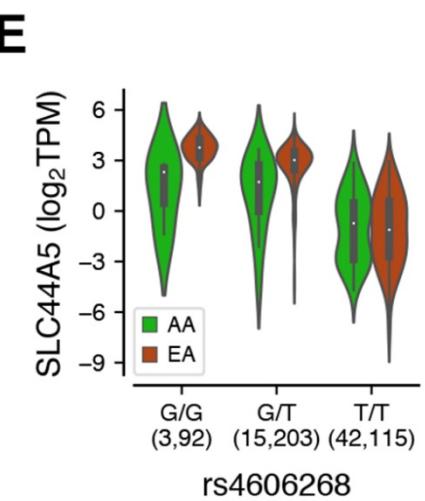
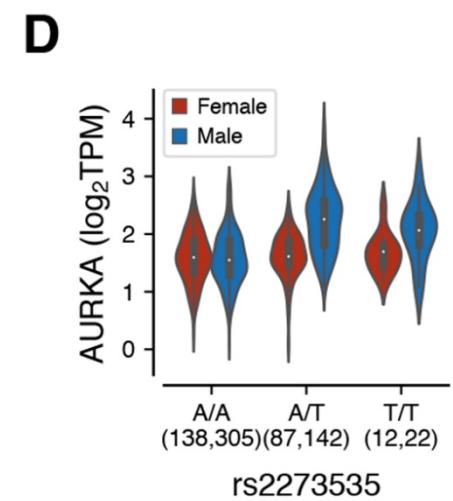
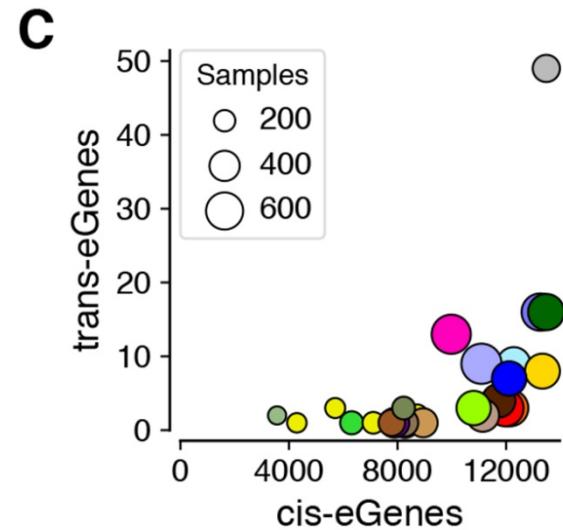
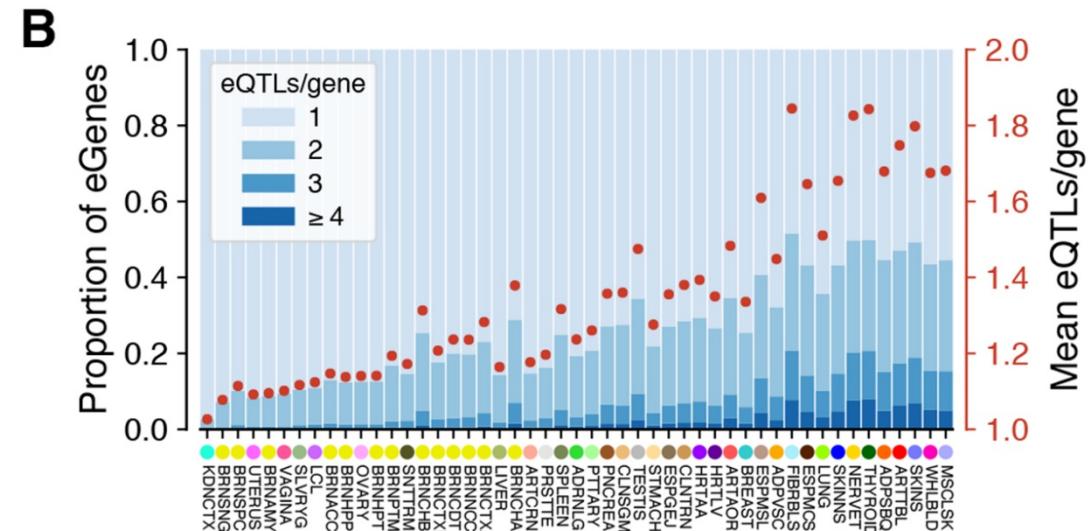
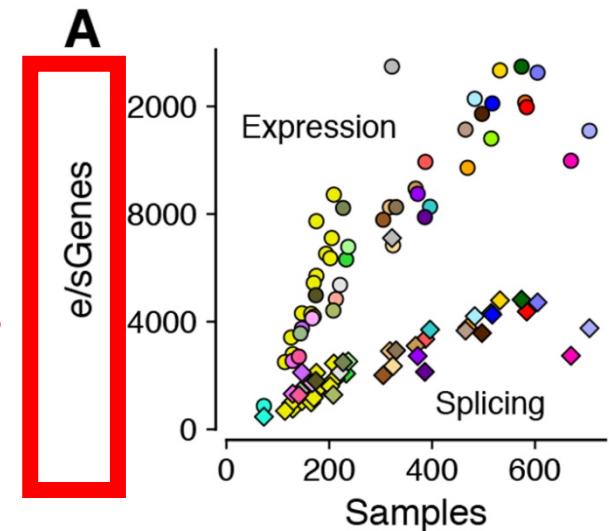


Fig2. QTL discovery

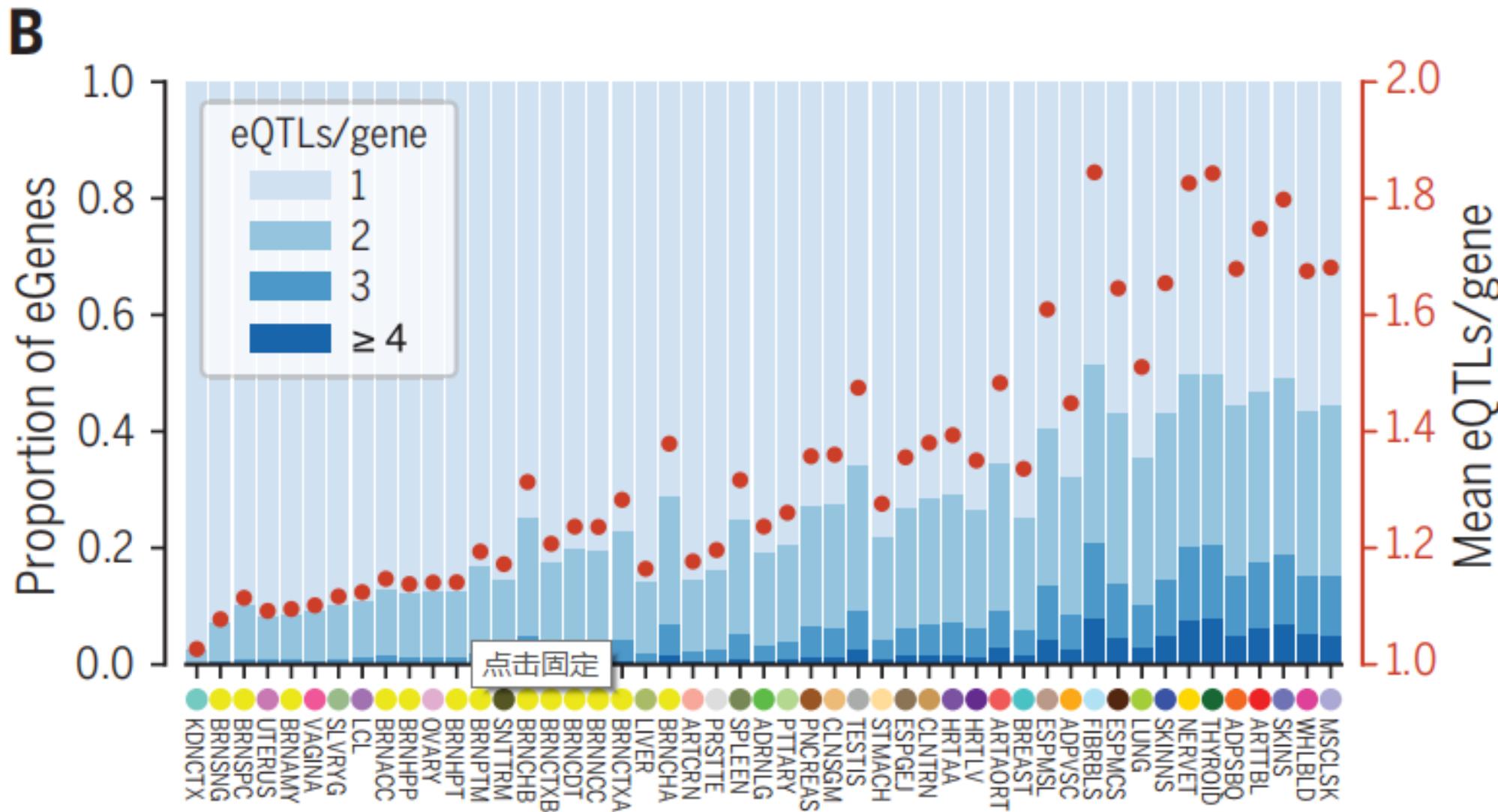


Fig2. QTL discovery

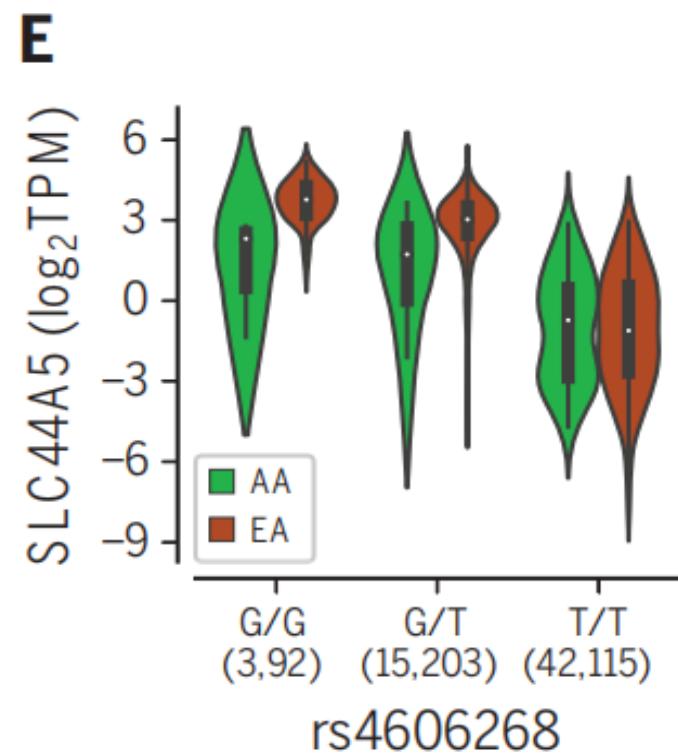
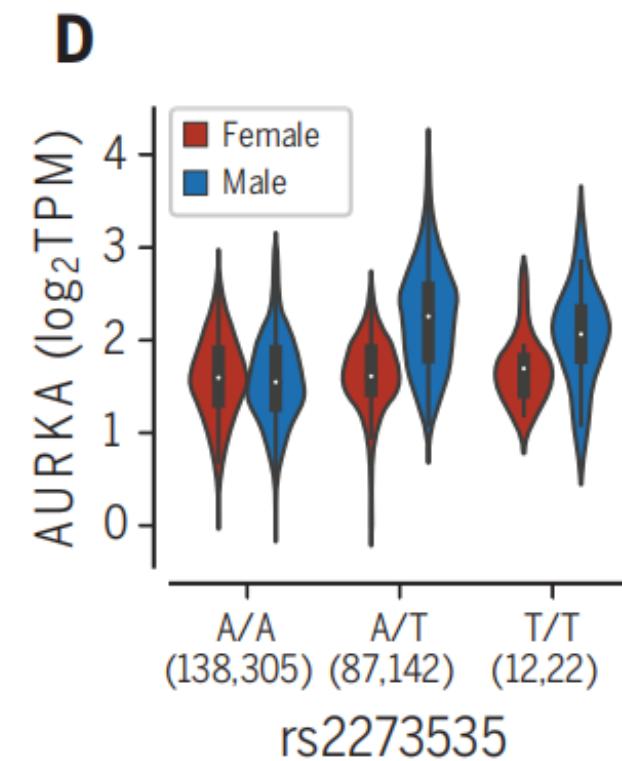
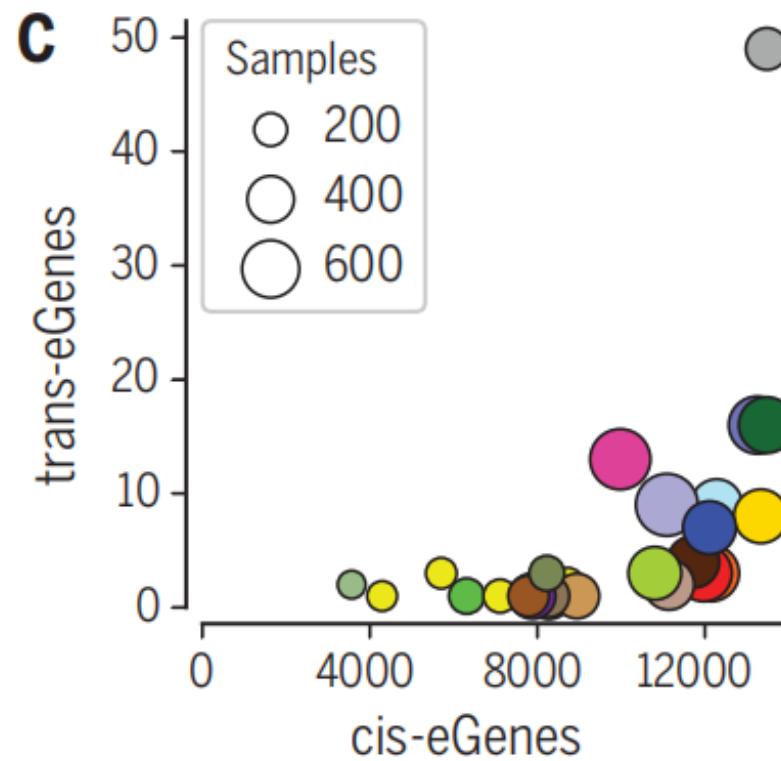


Fig3. Fine-mapping of cis-eQTLs

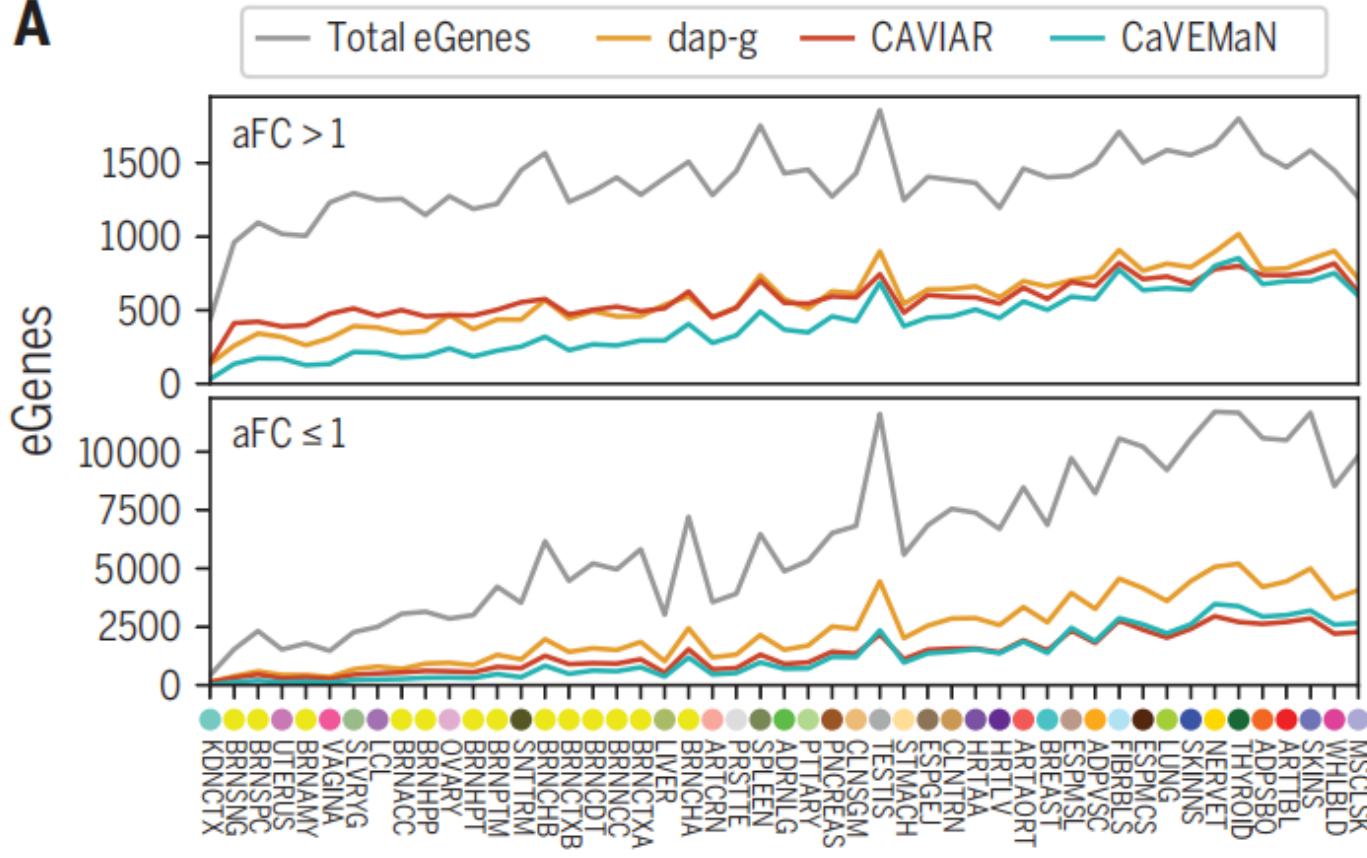
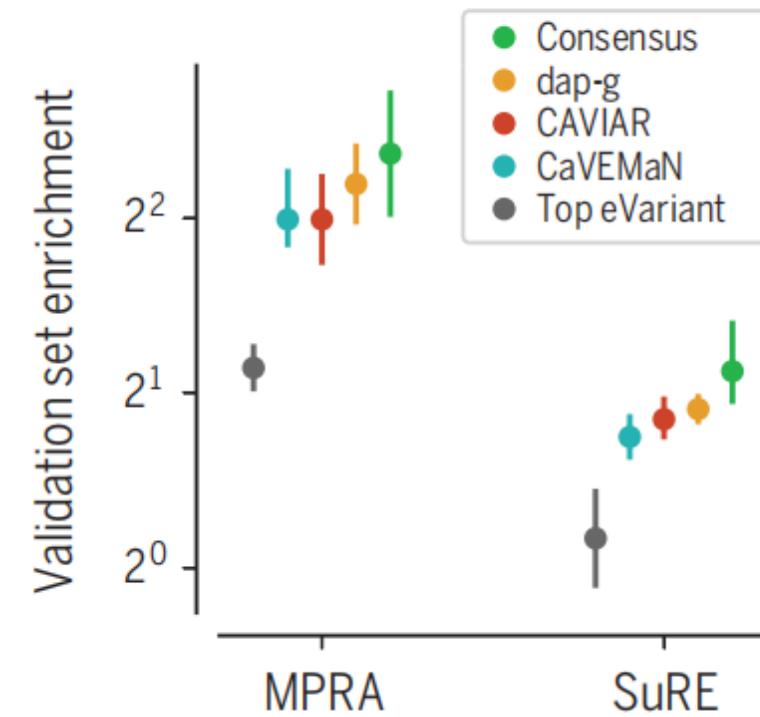
A**B**

Fig3. Fine-mapping of cis-eQTLs

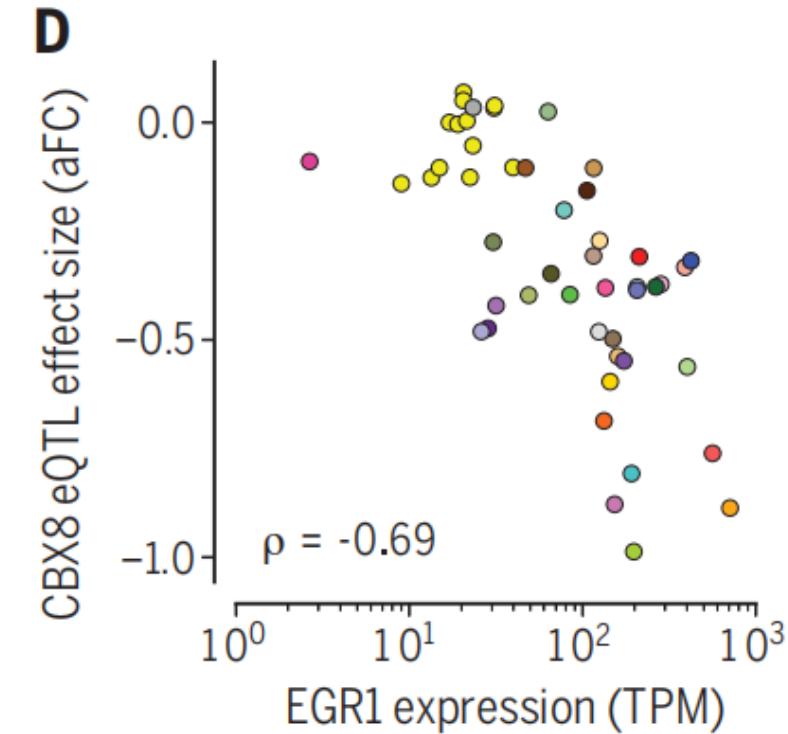
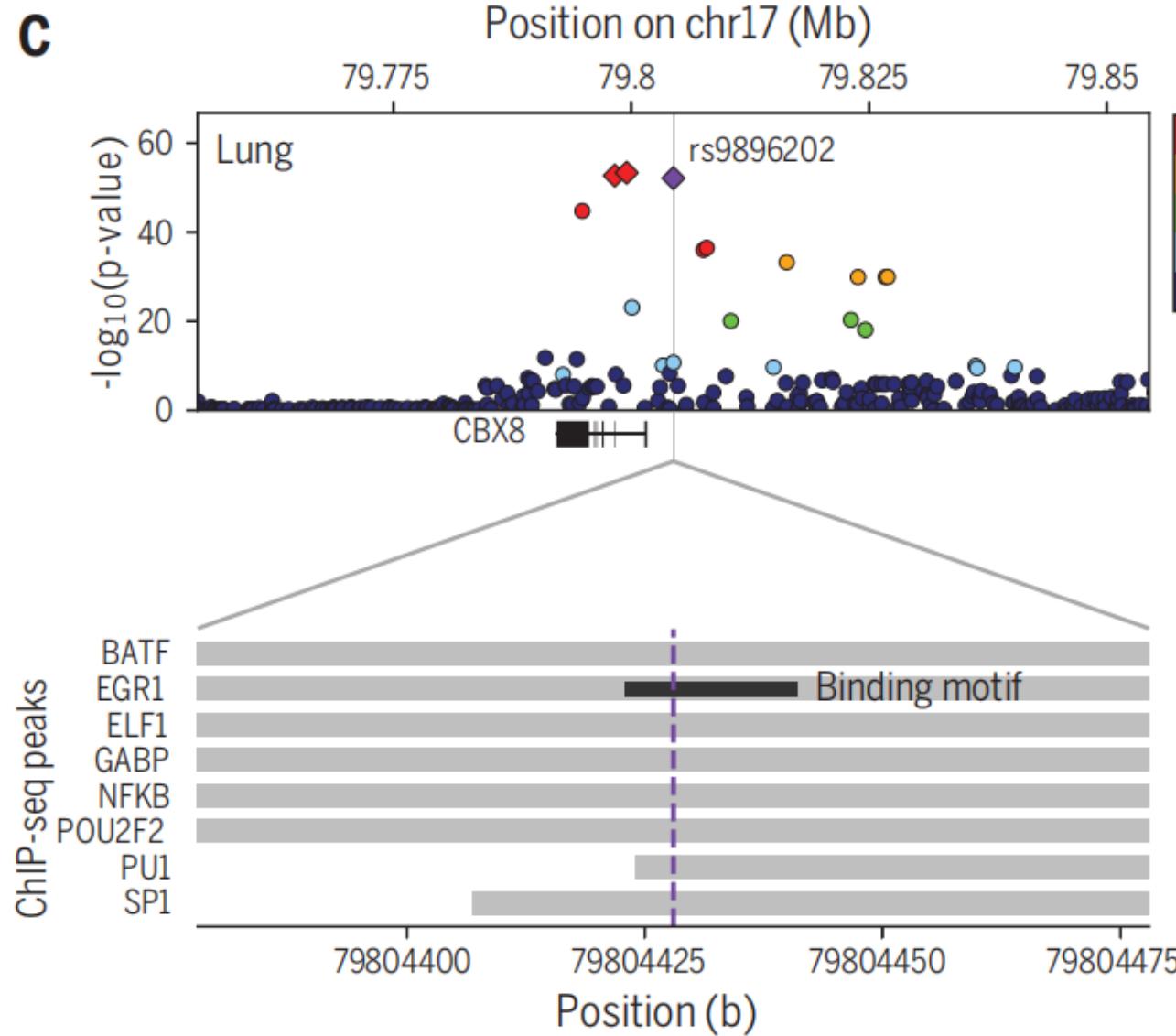


Fig4. Functional mechanisms of genetic regulatory effects

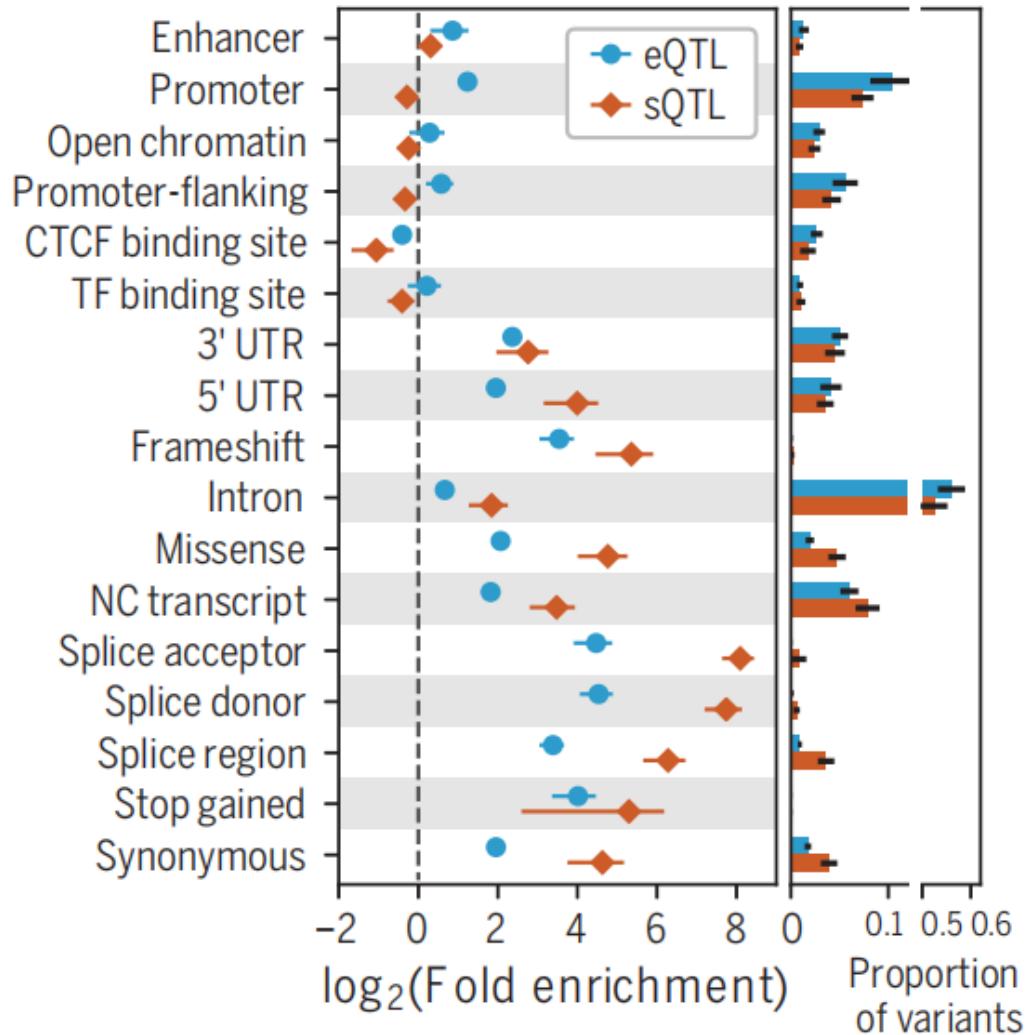
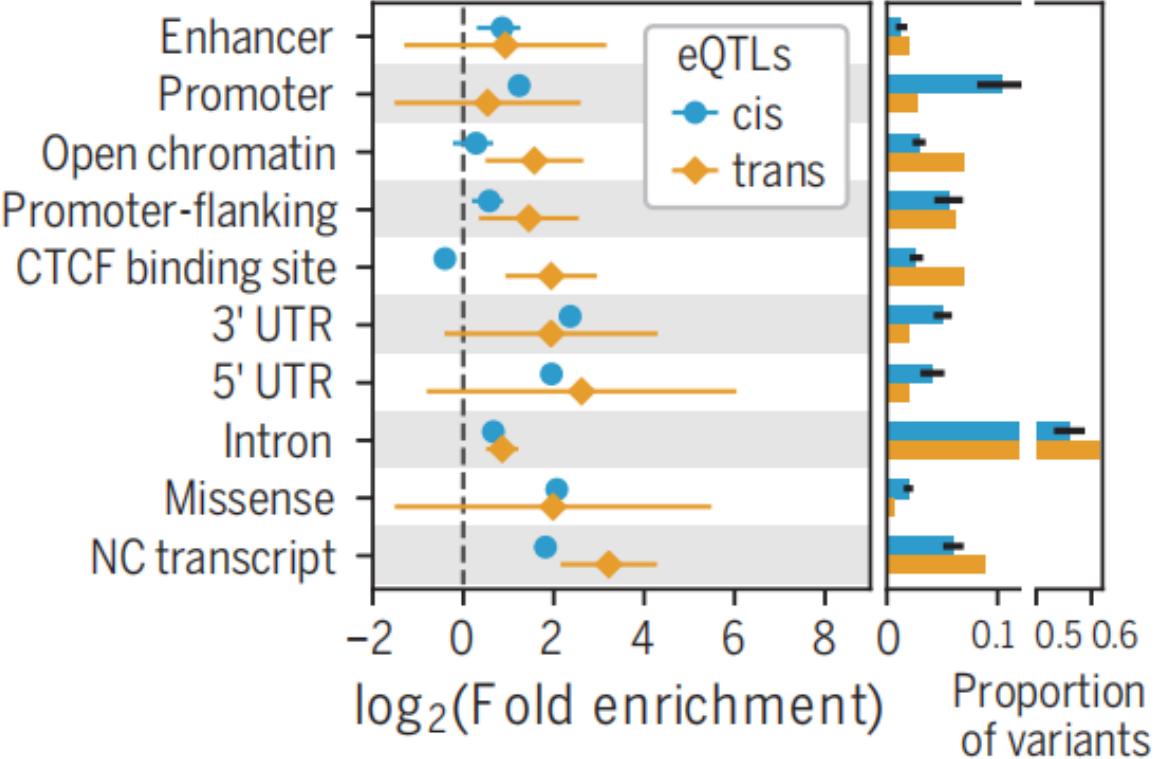
A**B**

Fig4. Functional mechanisms of genetic regulatory effects

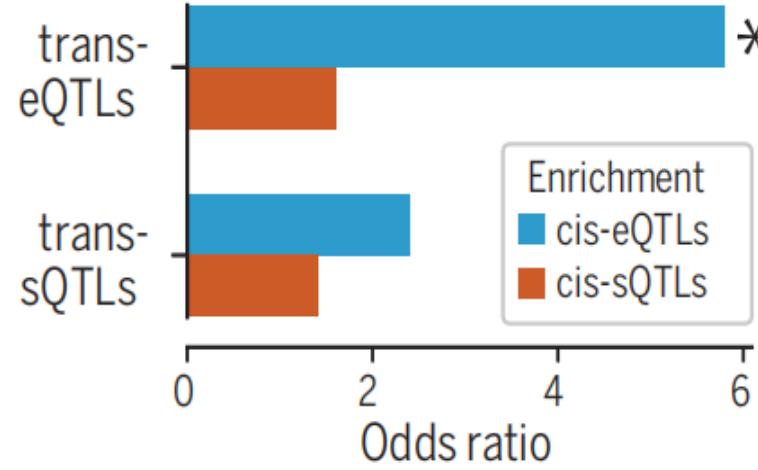
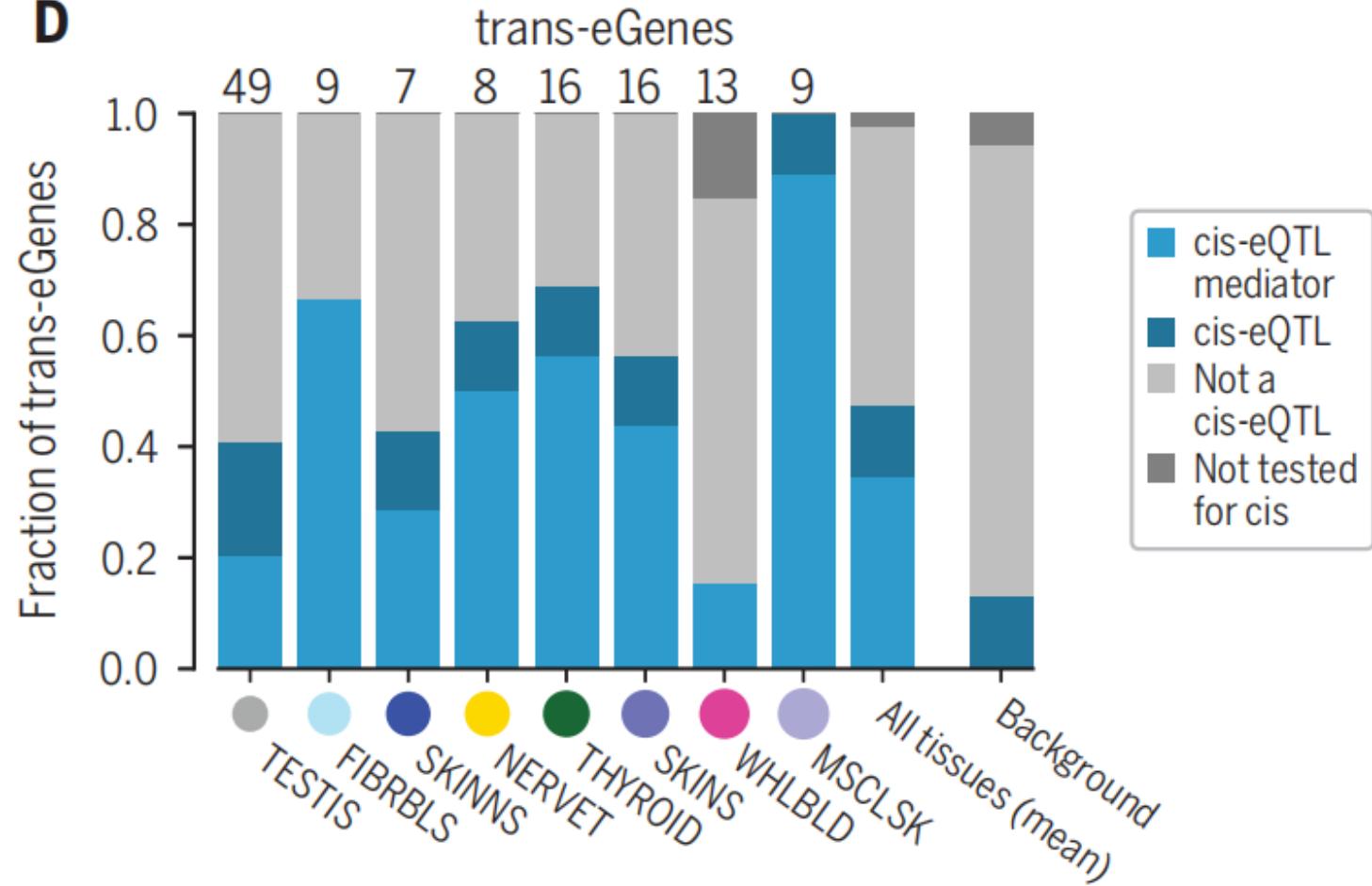
C**D**

Fig4. Functional mechanisms of genetic regulatory effects

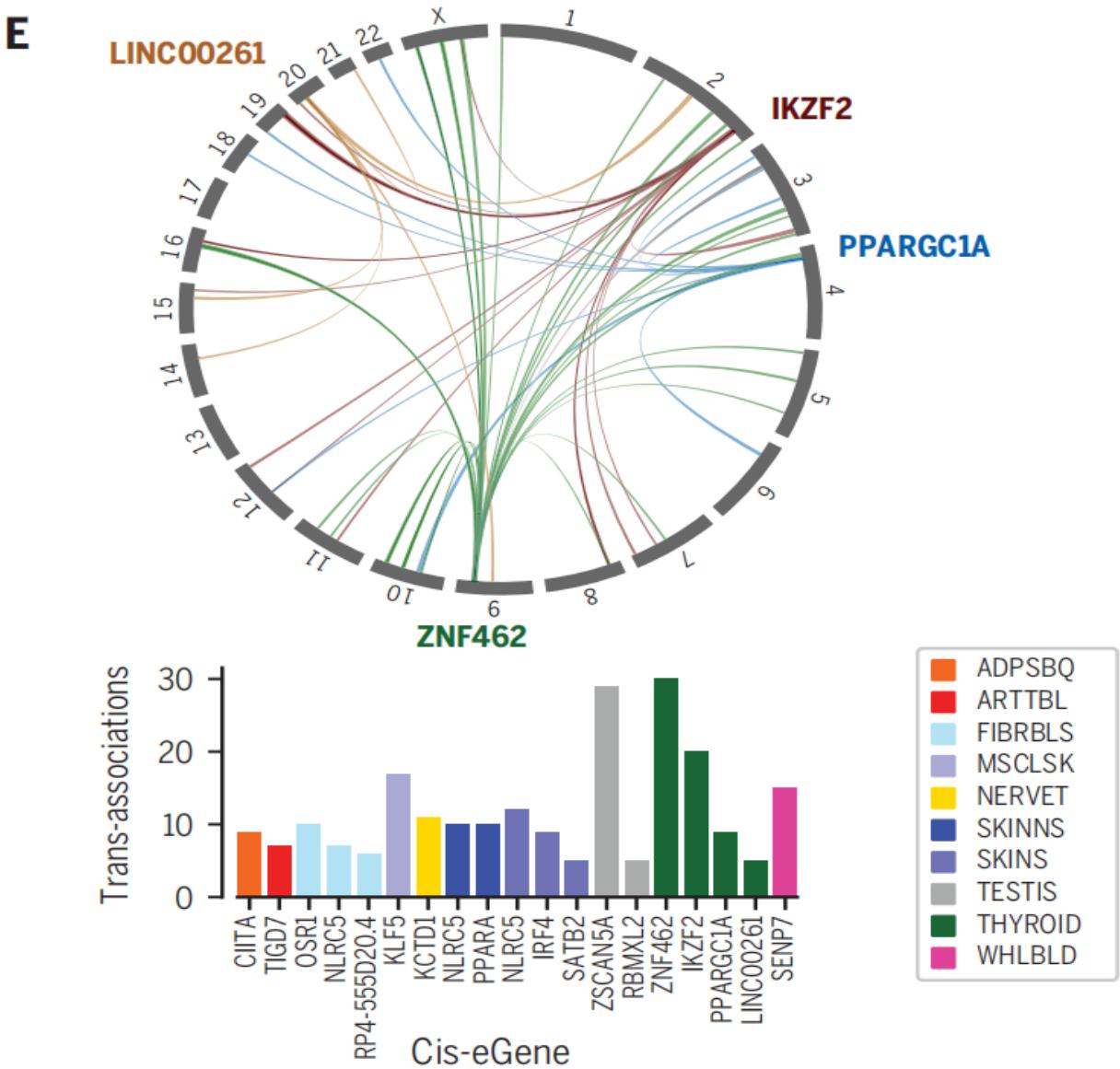


Fig5. Regulatory mechanisms of GWAS loci

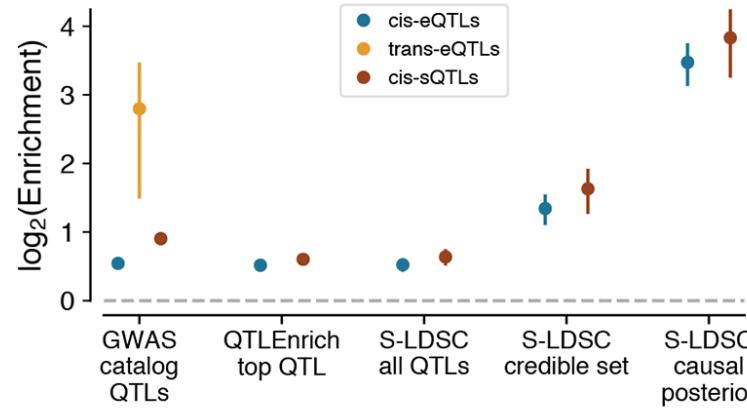
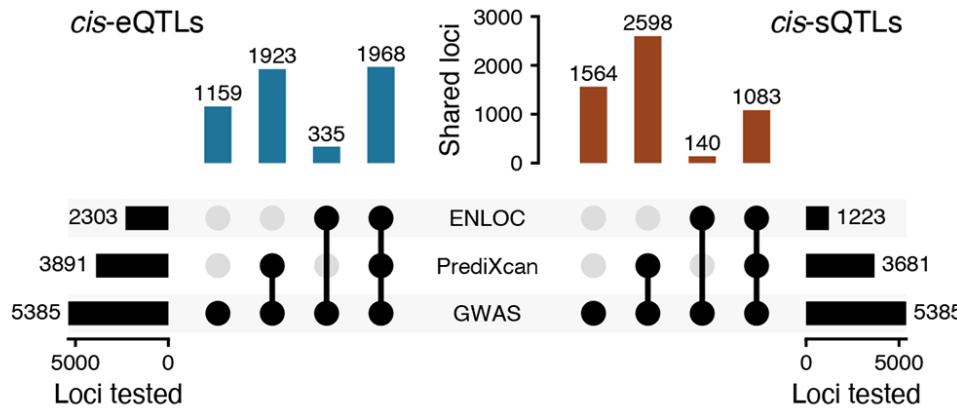
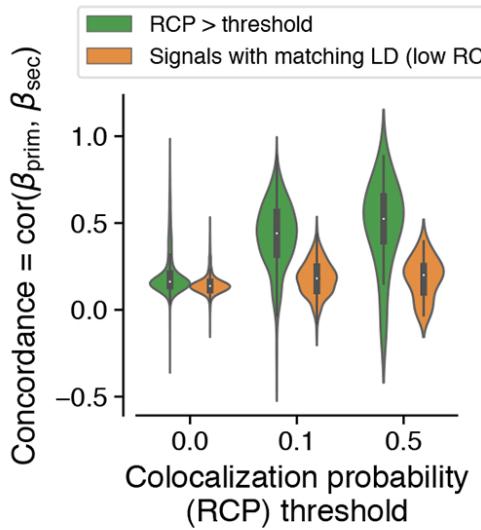
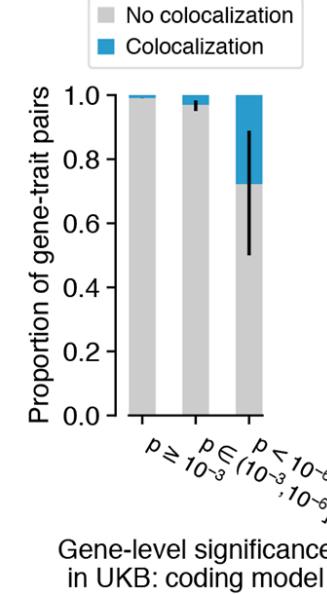
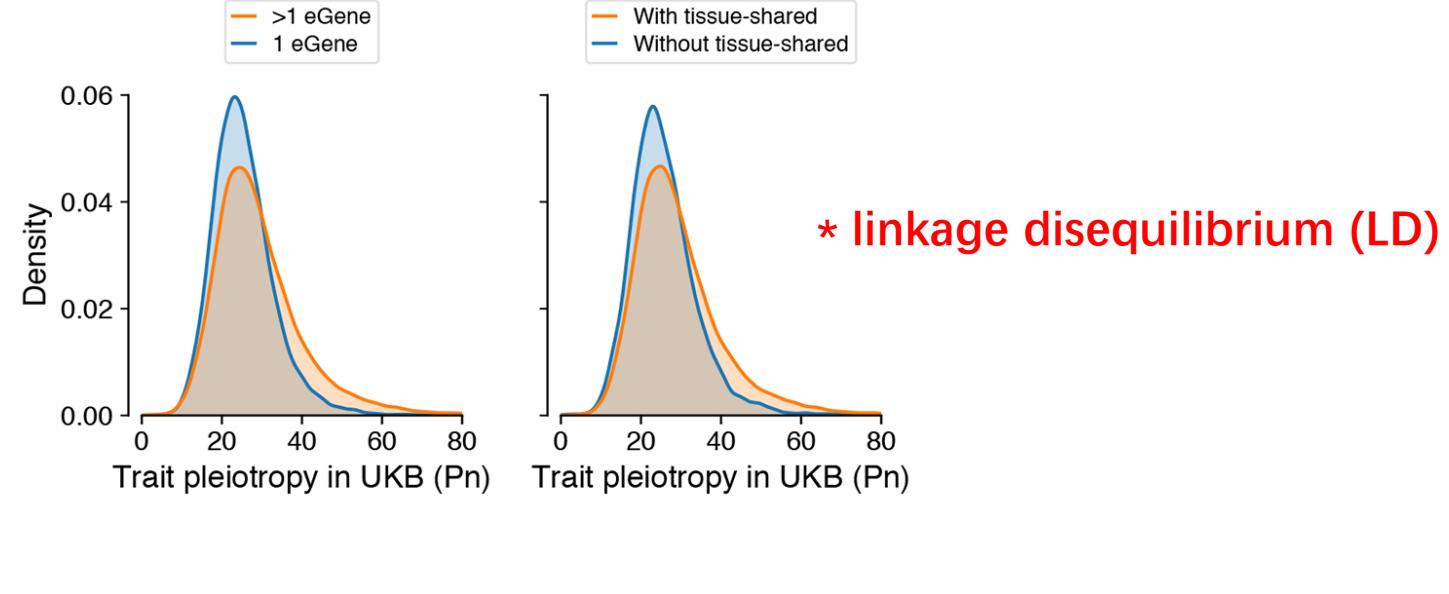
A**B****C****D****E**

Fig5. Regulatory mechanisms of GWAS loci

GWAS位点的调控机制

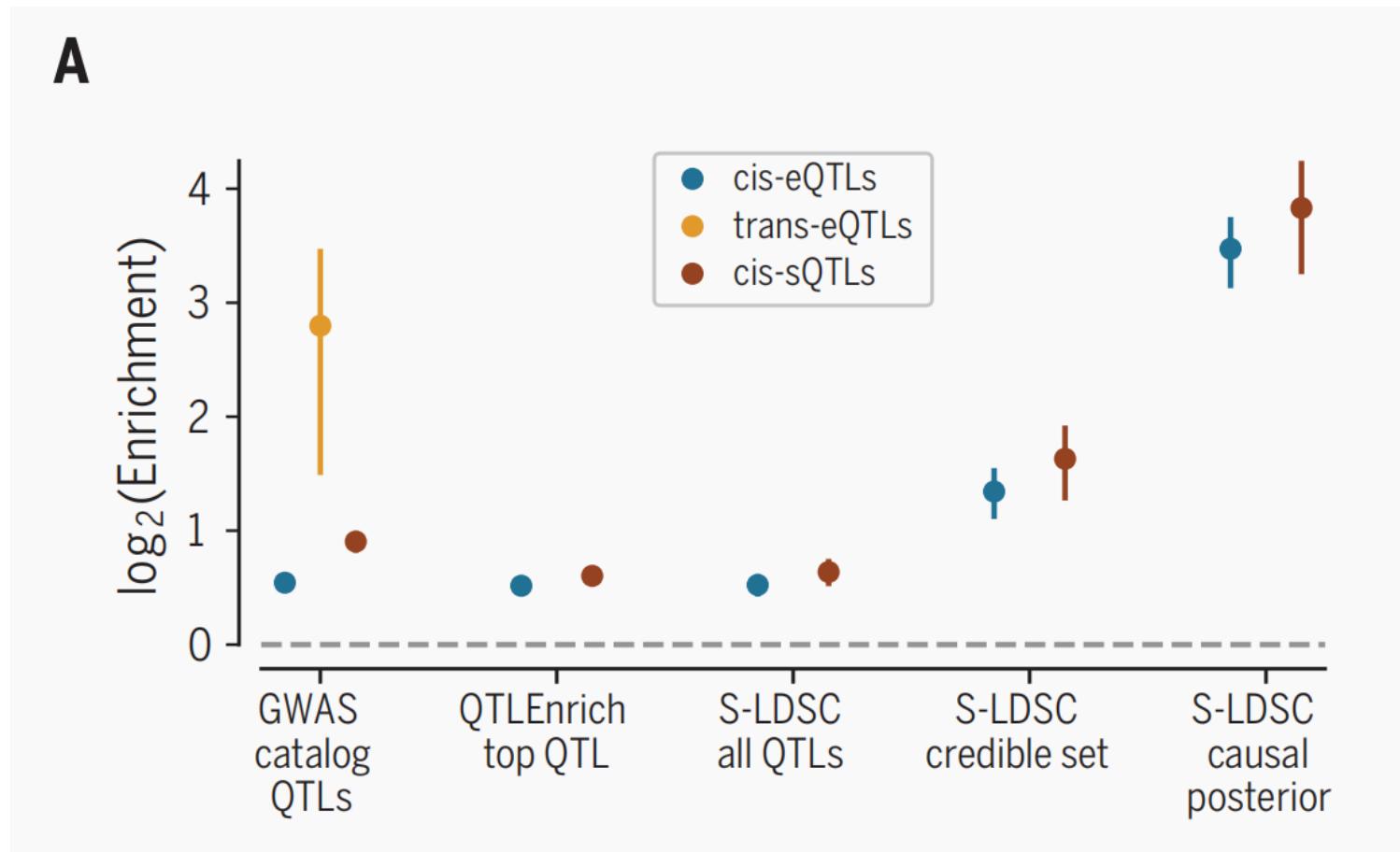


Fig5. Regulatory mechanisms of GWAS loci

聚集在组织中的通过共定位(ENLOC)和关联(PrediXcan)与eGenes或者sGenes相连的GWAS位点数量

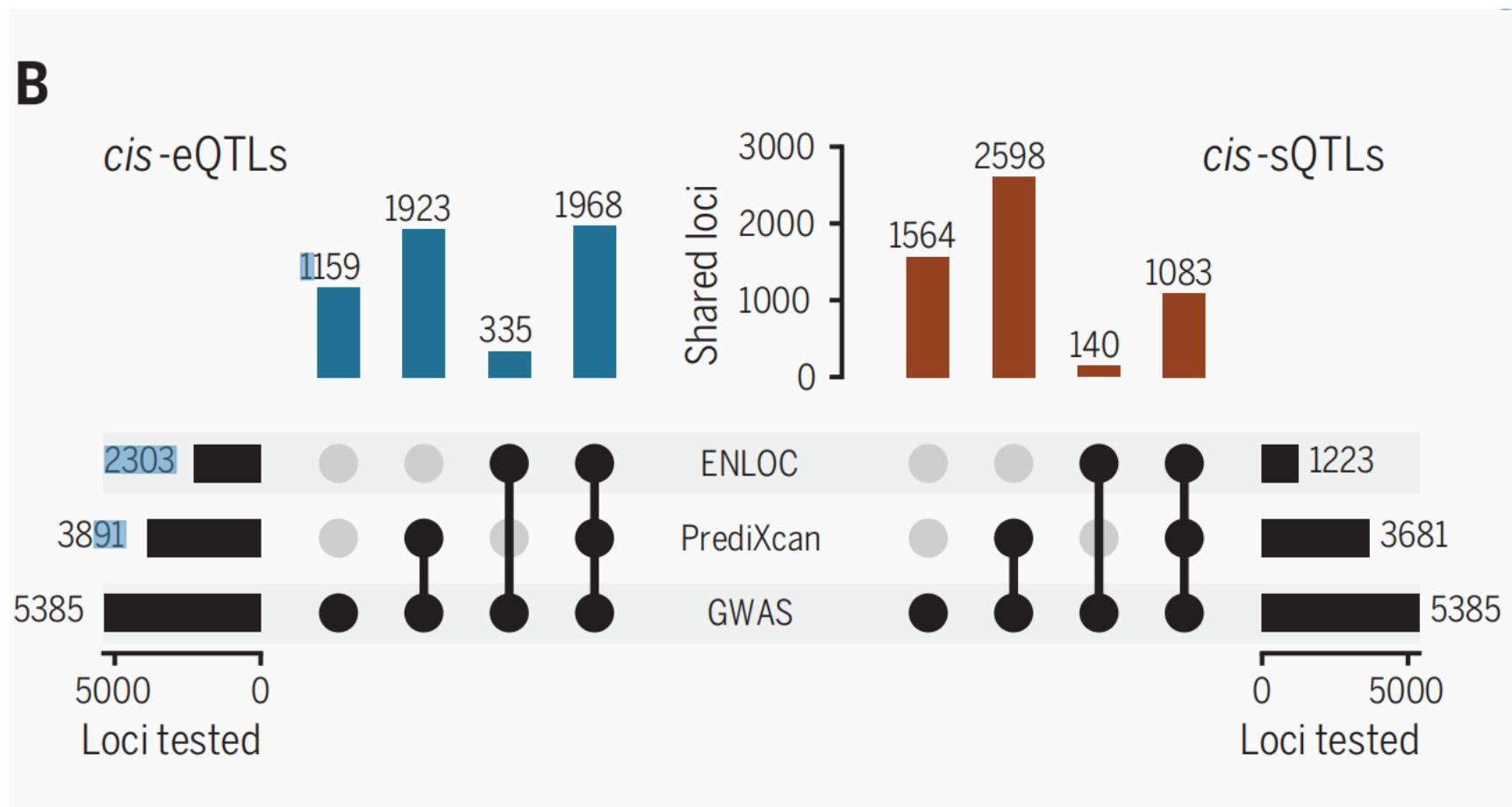


Fig5. Regulatory mechanisms of GWAS loci

在同一基因的独立顺式-eQTL之间的介导效应的一致性，显示出不同水平的区域共定位概率(RCP)

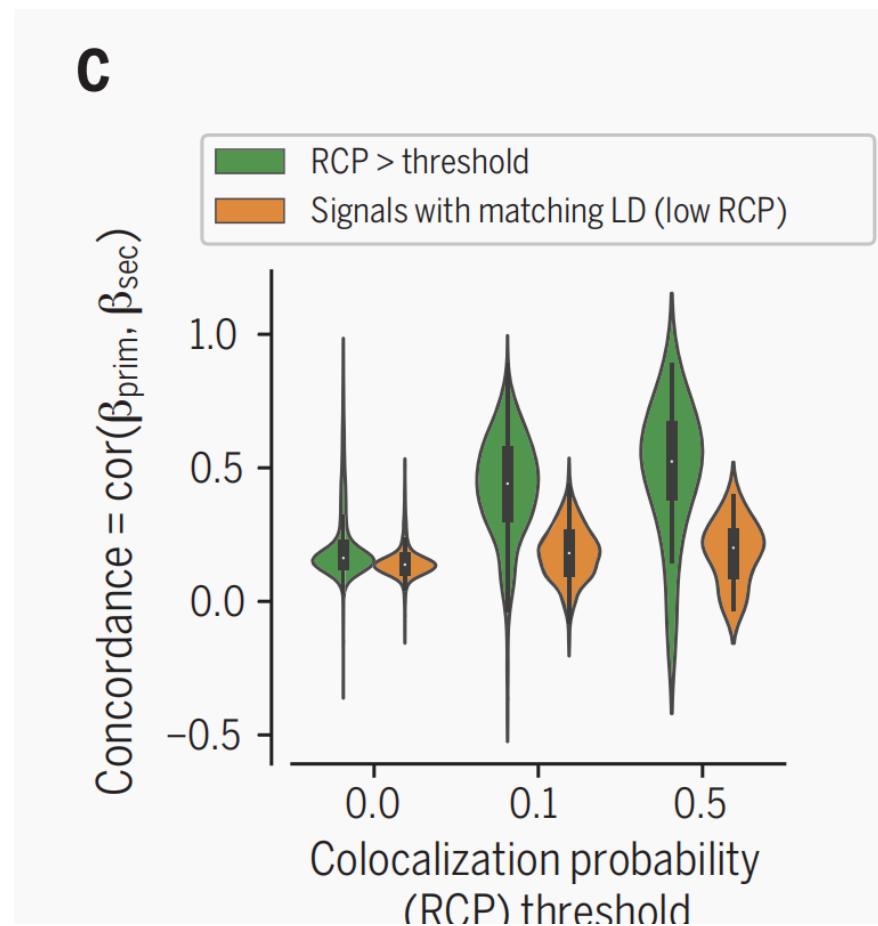


Fig5. Regulatory mechanisms of GWAS loci

UKB中具有不同水平罕见变异性状关联的基因具有匹配表型的共定位顺式eQTLs的比例

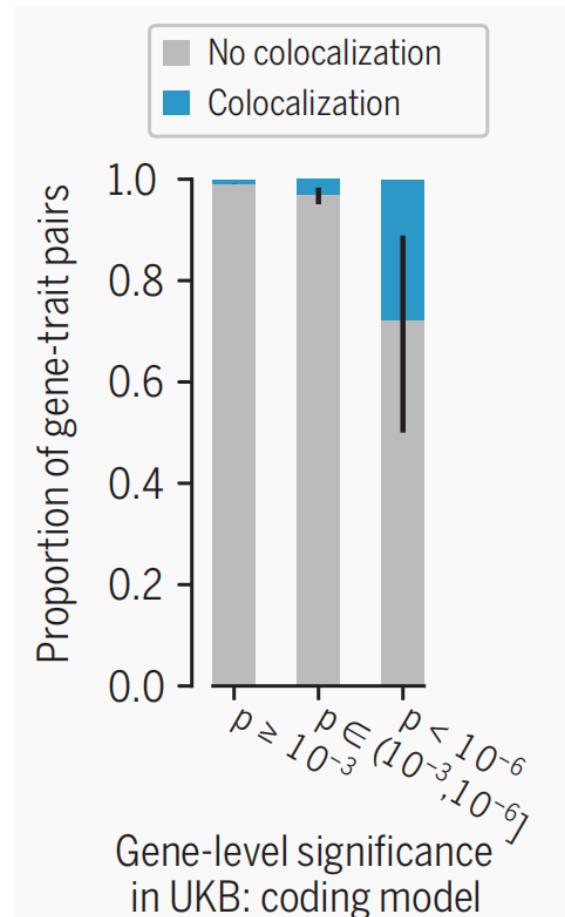


Fig6. Tissue-specificity of cis-QTLs

顺式-eQTLs效应大小的两两斯皮尔曼相关性的组织聚类

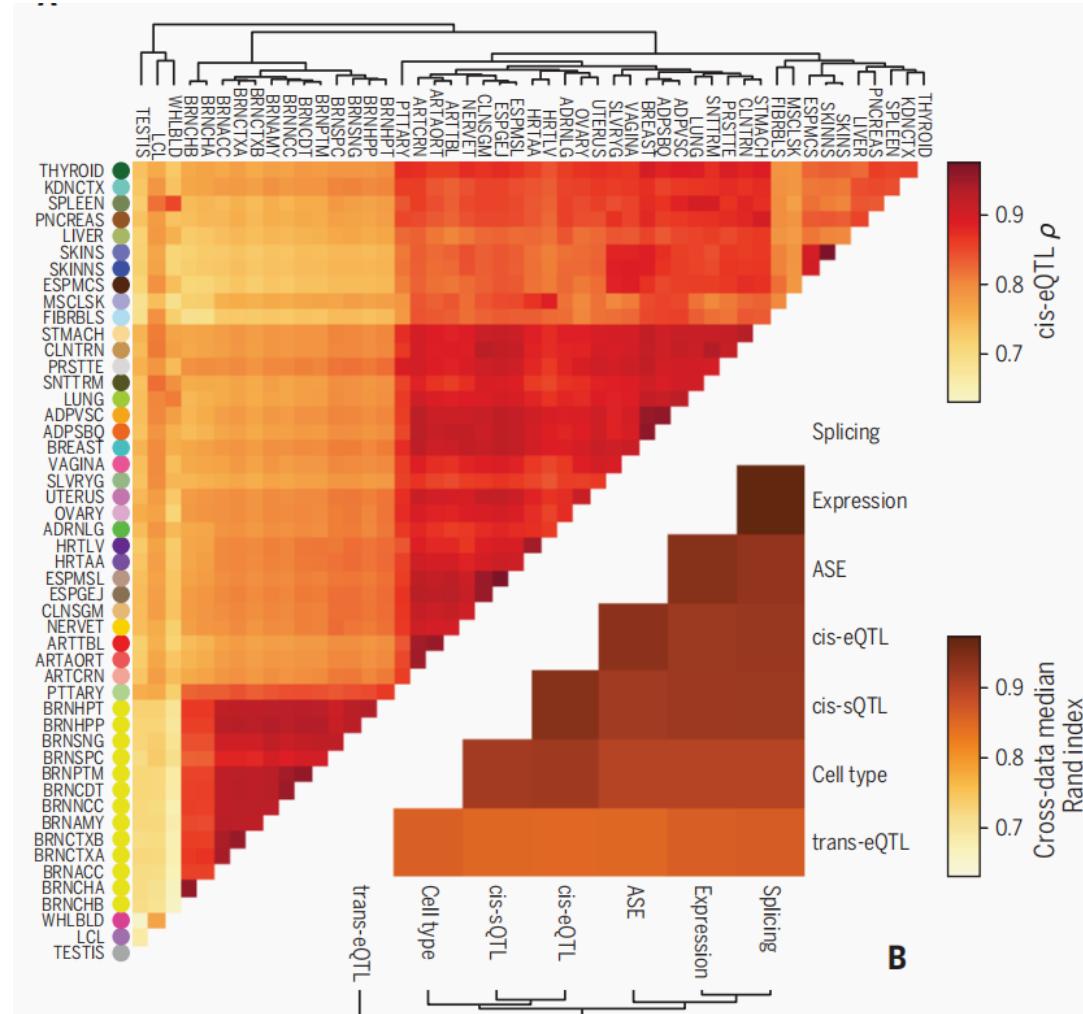


Fig6. Tissue-specificity of cis-QTLs

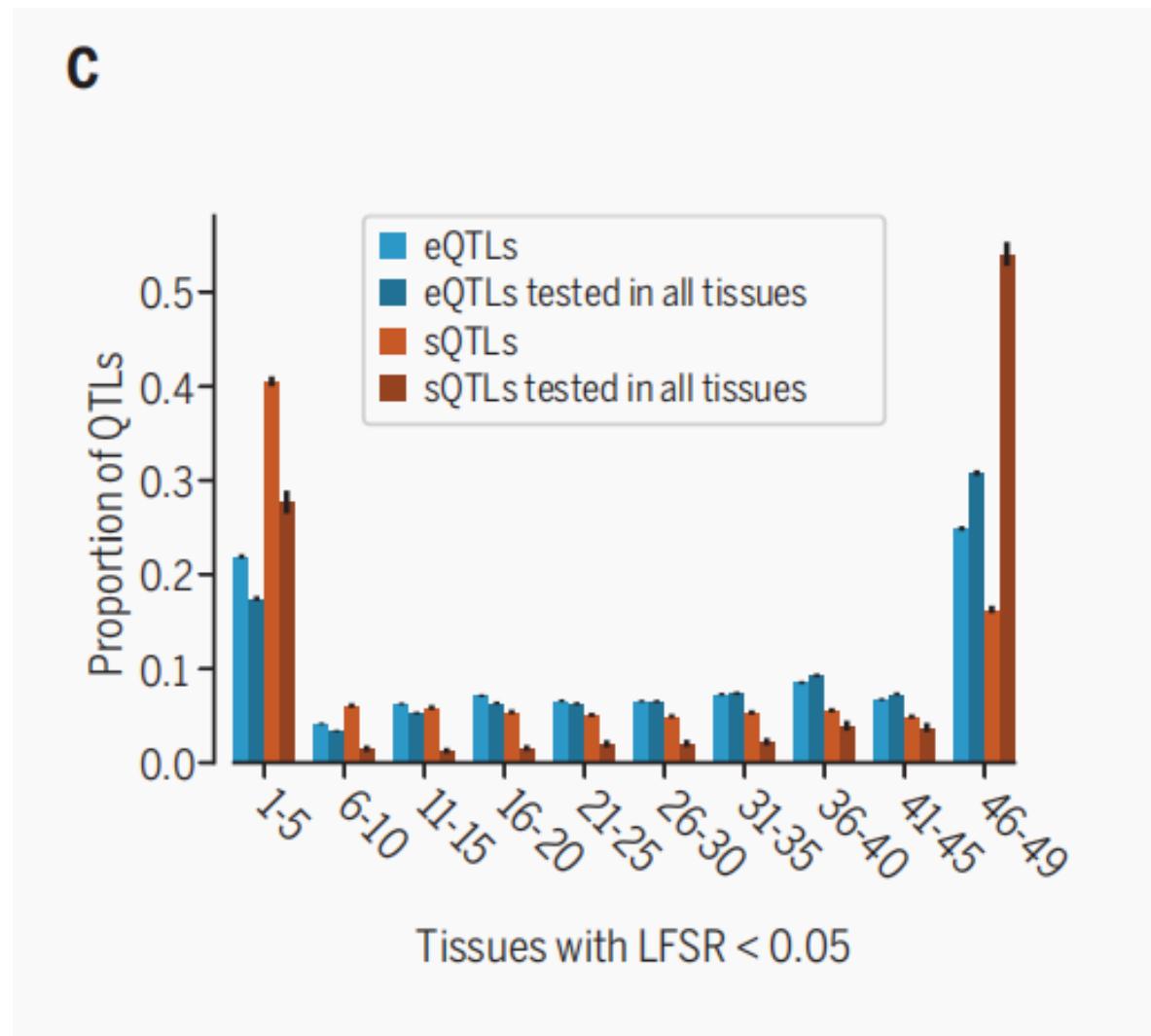


Fig6. Tissue-specificity of cis-QTLs

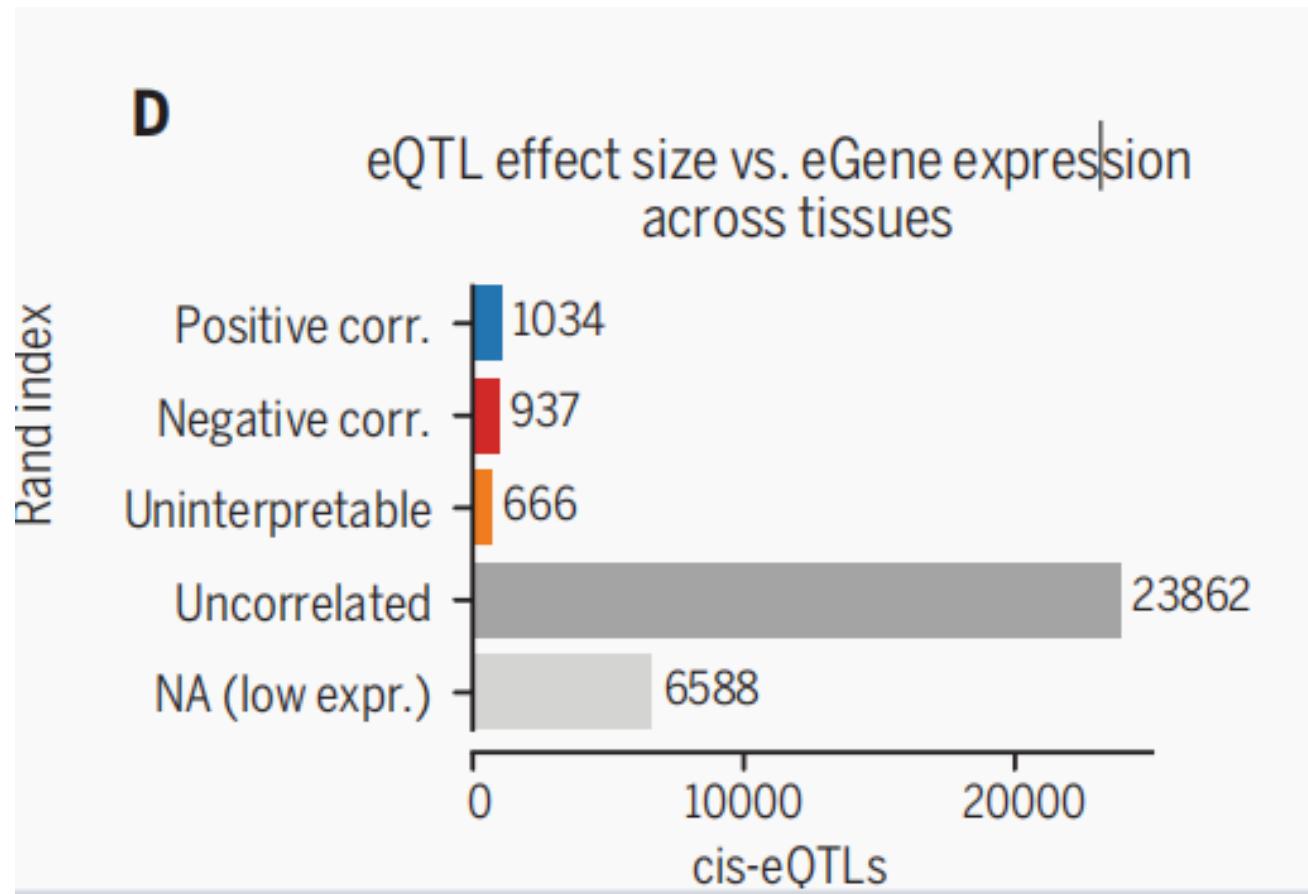


Fig6. Tissue-specificity of cis-QTLs

基因组功能对使用带有功能注释的逻辑回归建立顺式-QTL组织共享模型的和染色质状态的影响

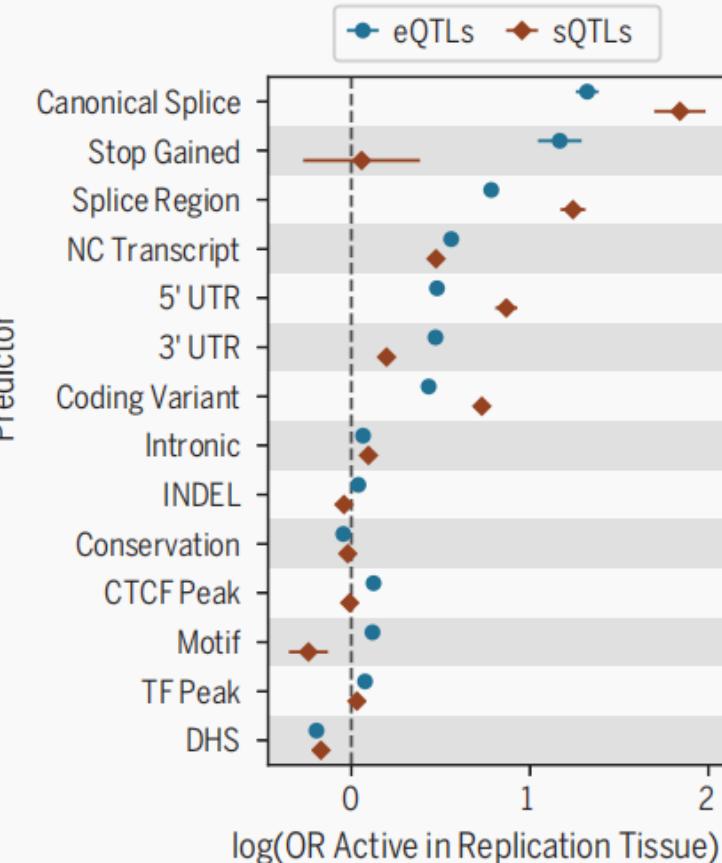
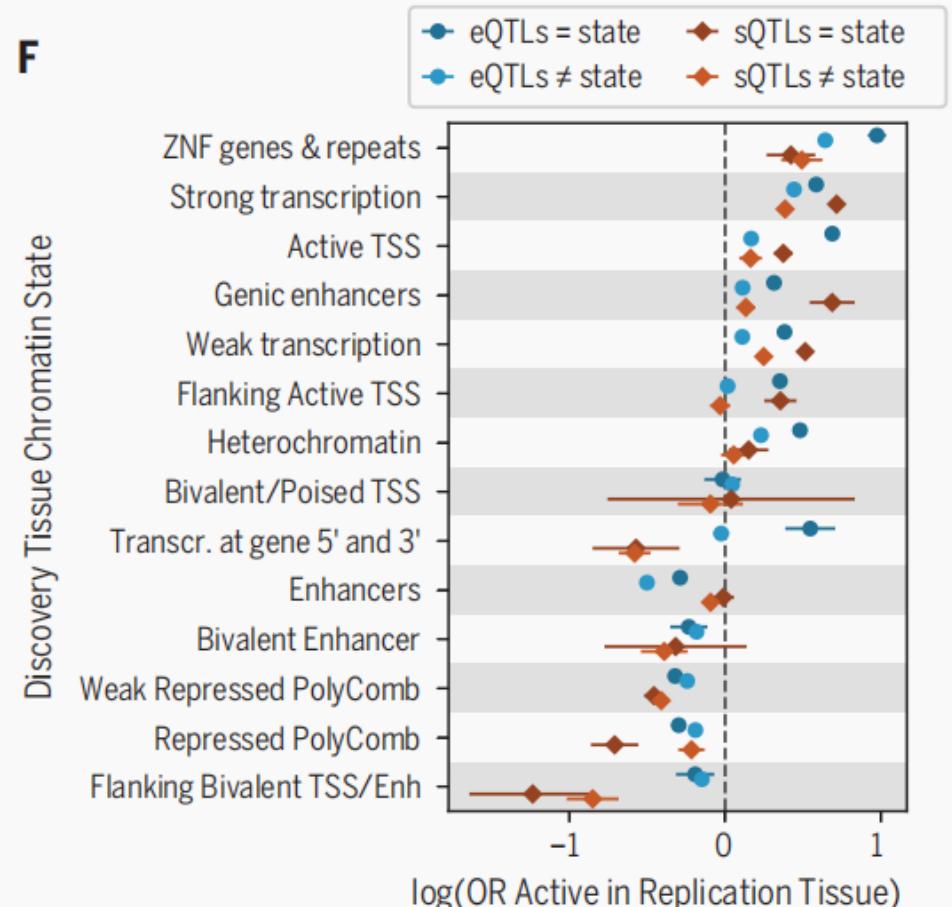
E**F**

Fig7. Cell type interacting cis-eQTLs and cis-sQTLs

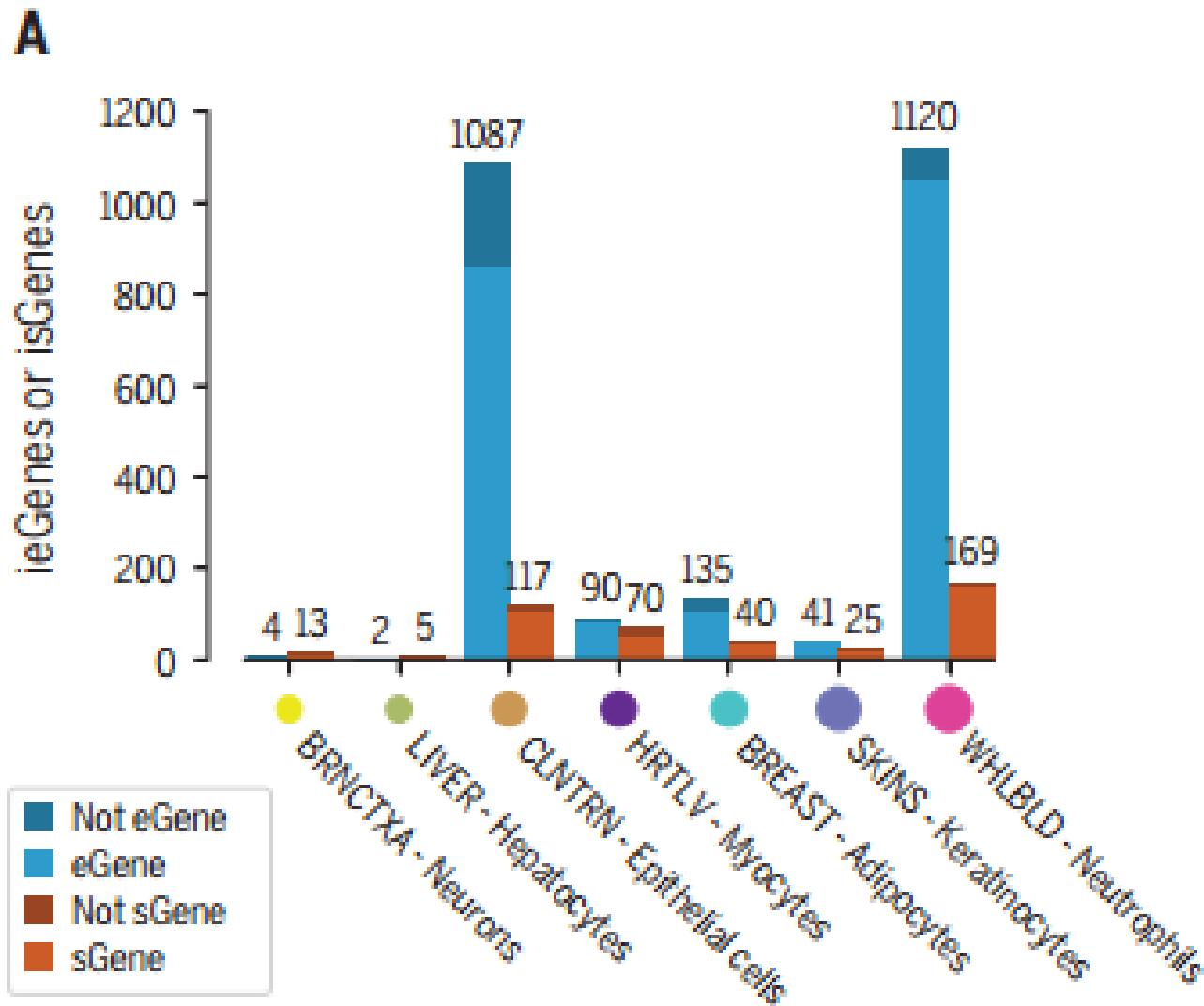


Fig7. Cell type interacting cis-eQTLs and cis-sQTLs

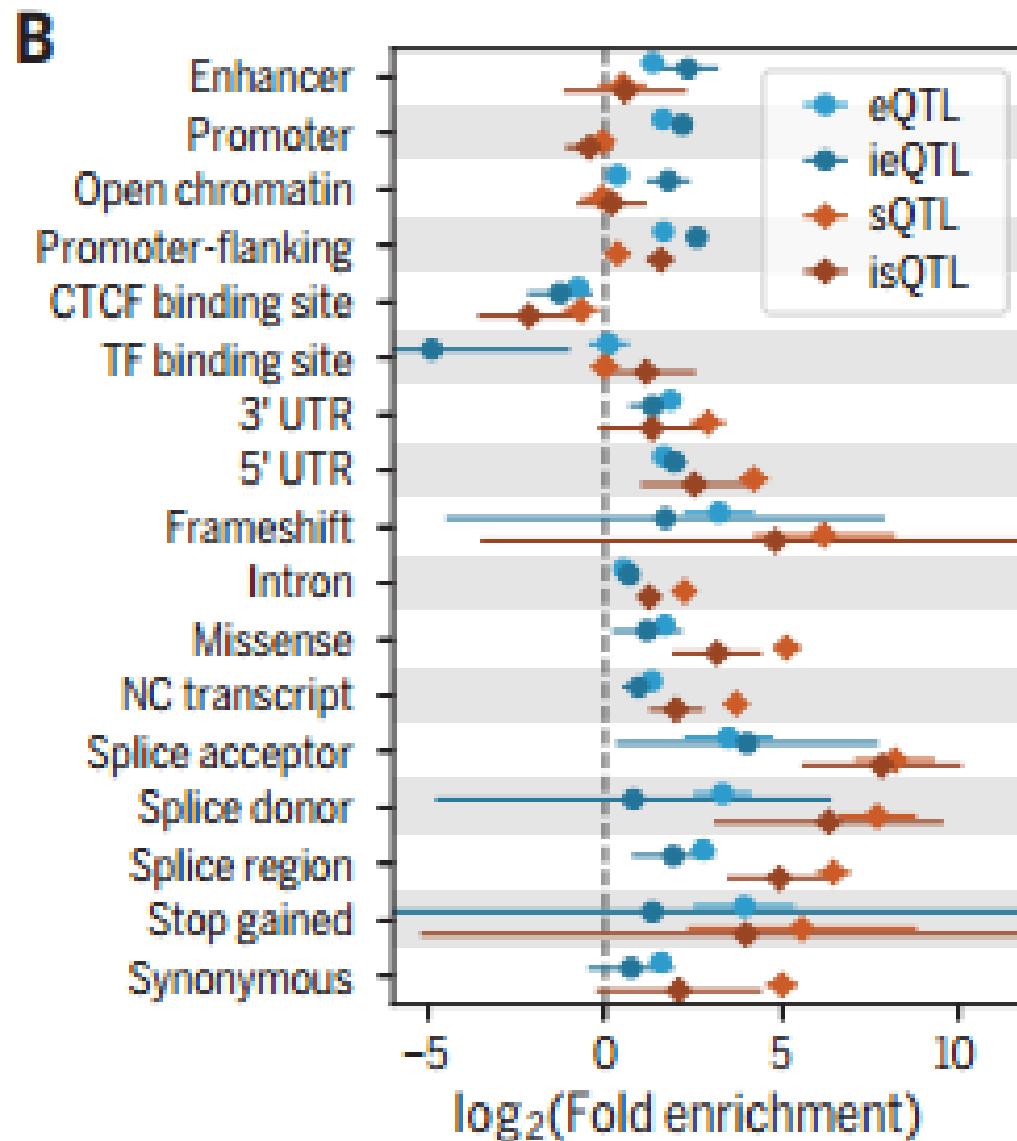


Fig7. Cell type interacting cis-eQTLs and cis-sQTLs

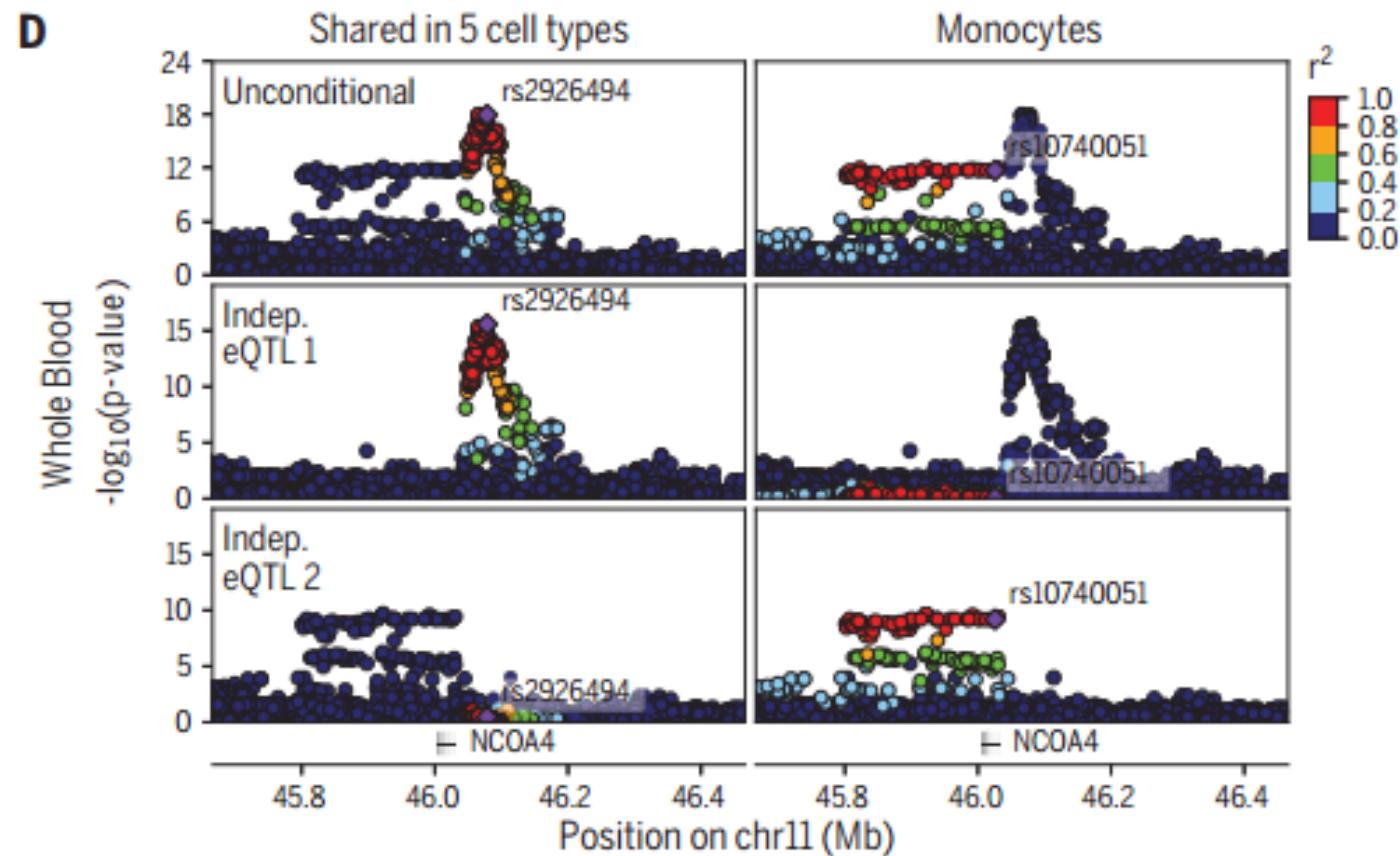
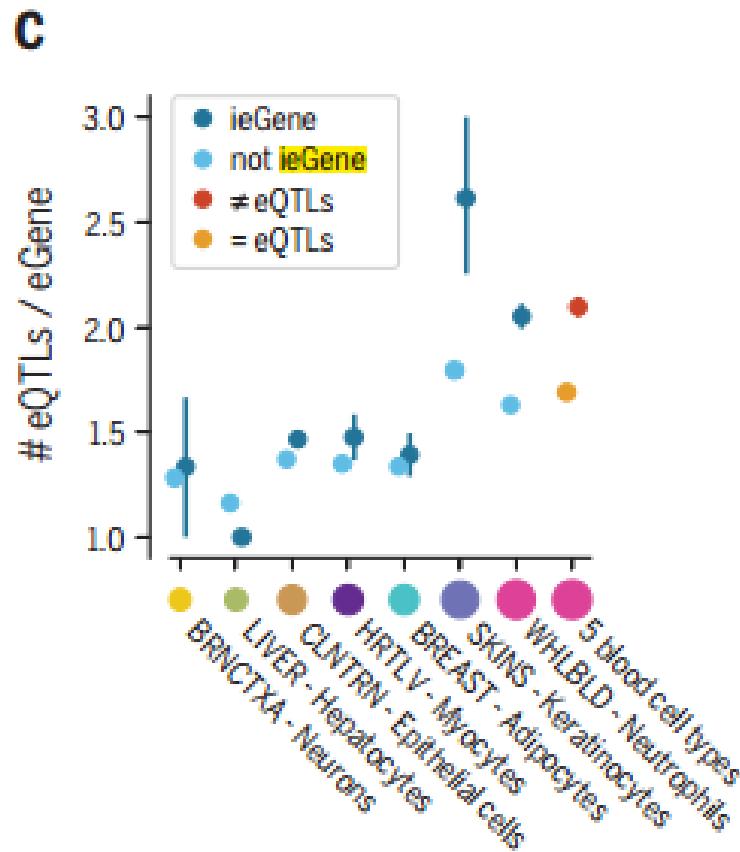
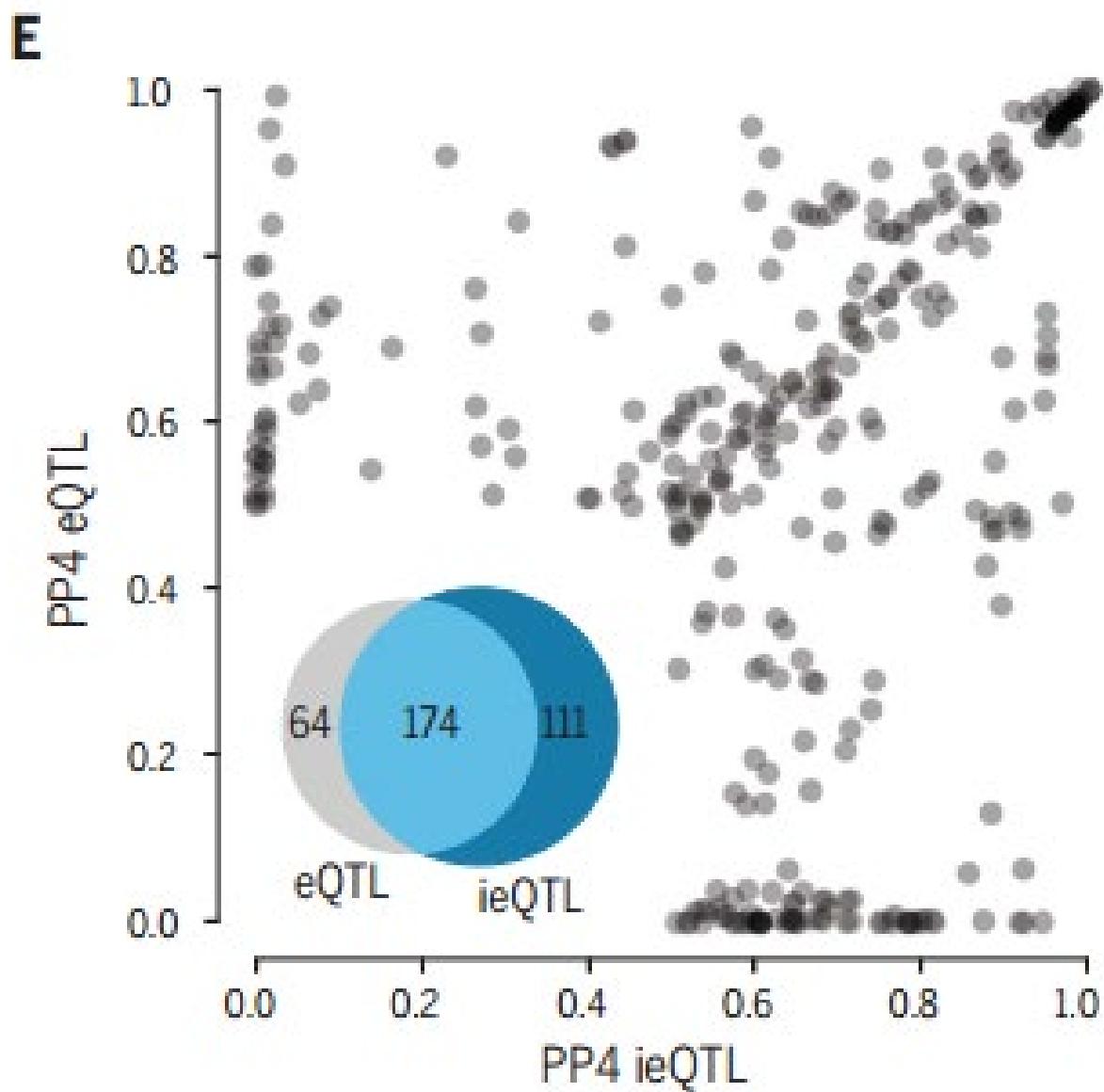
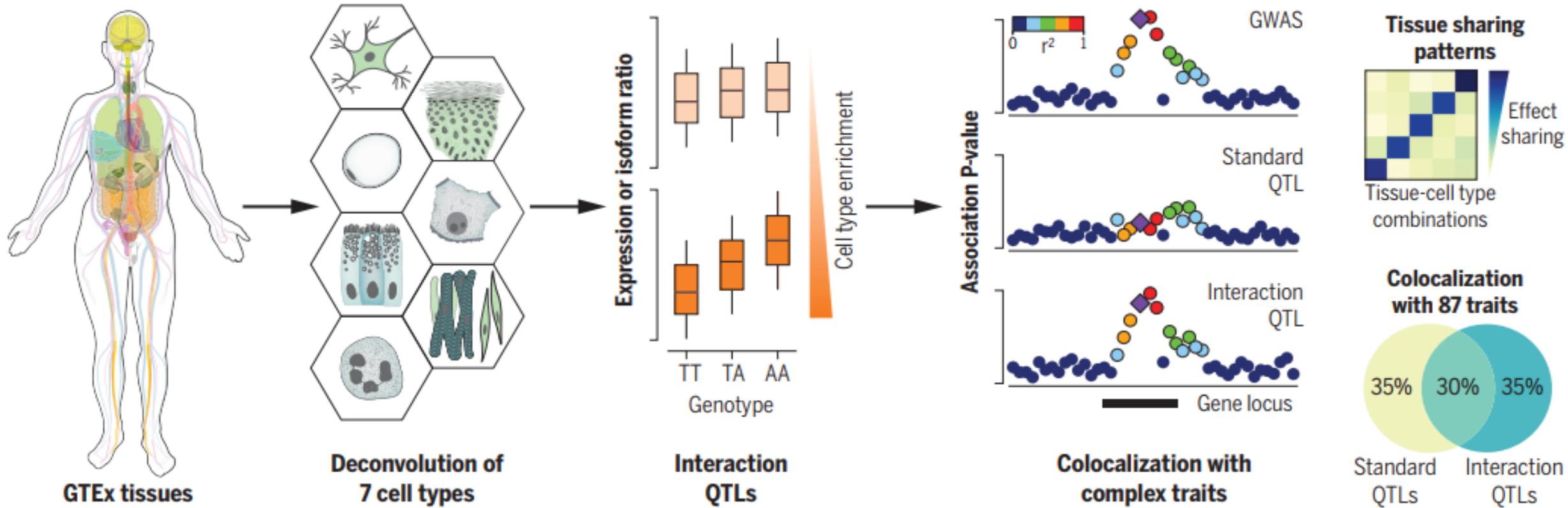


Fig7. Cell type interacting cis-eQTLs and cis-sQTLs



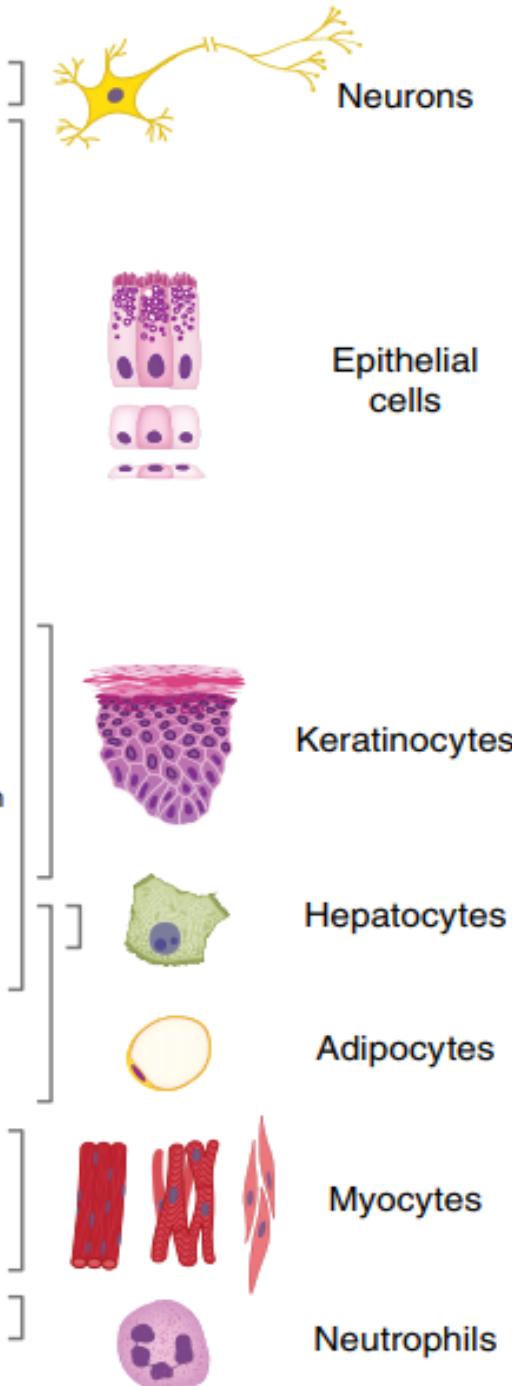
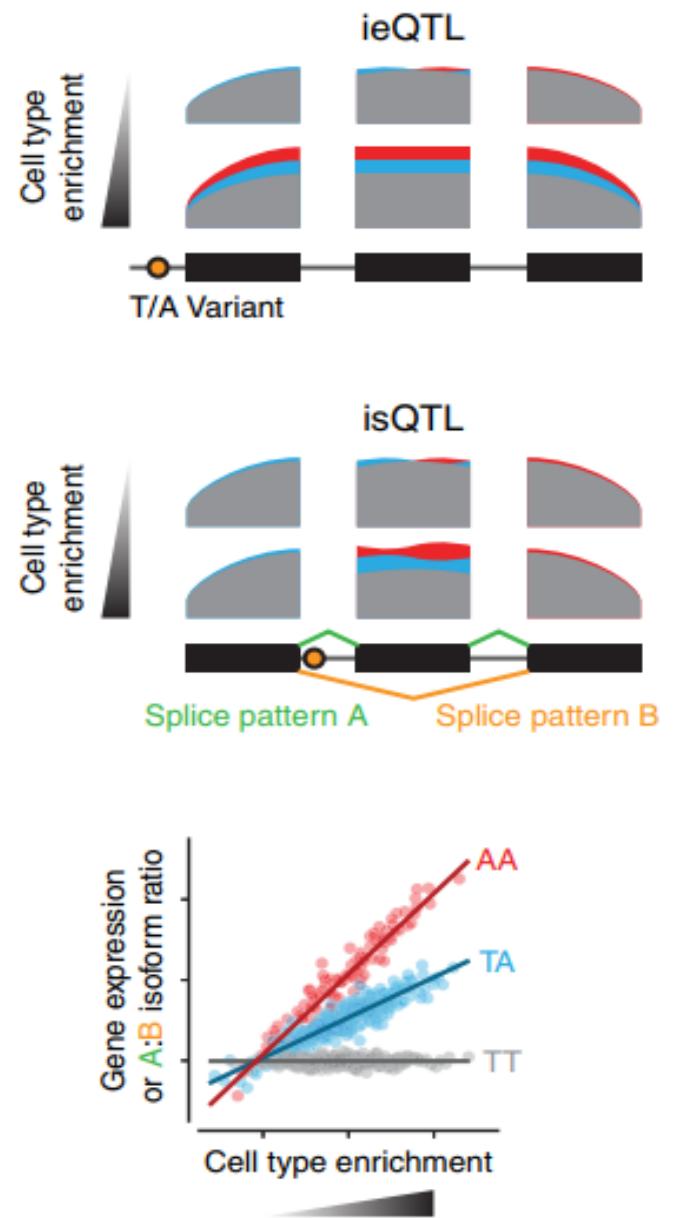
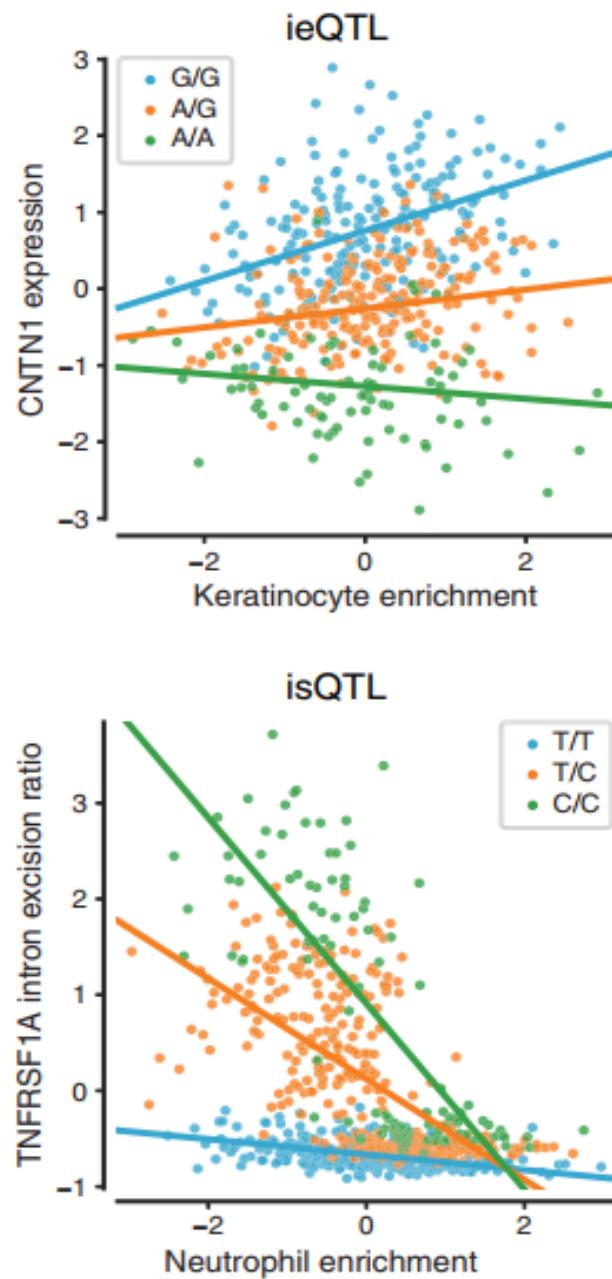
Cell type specific genetic regulation of gene expression across human tissues

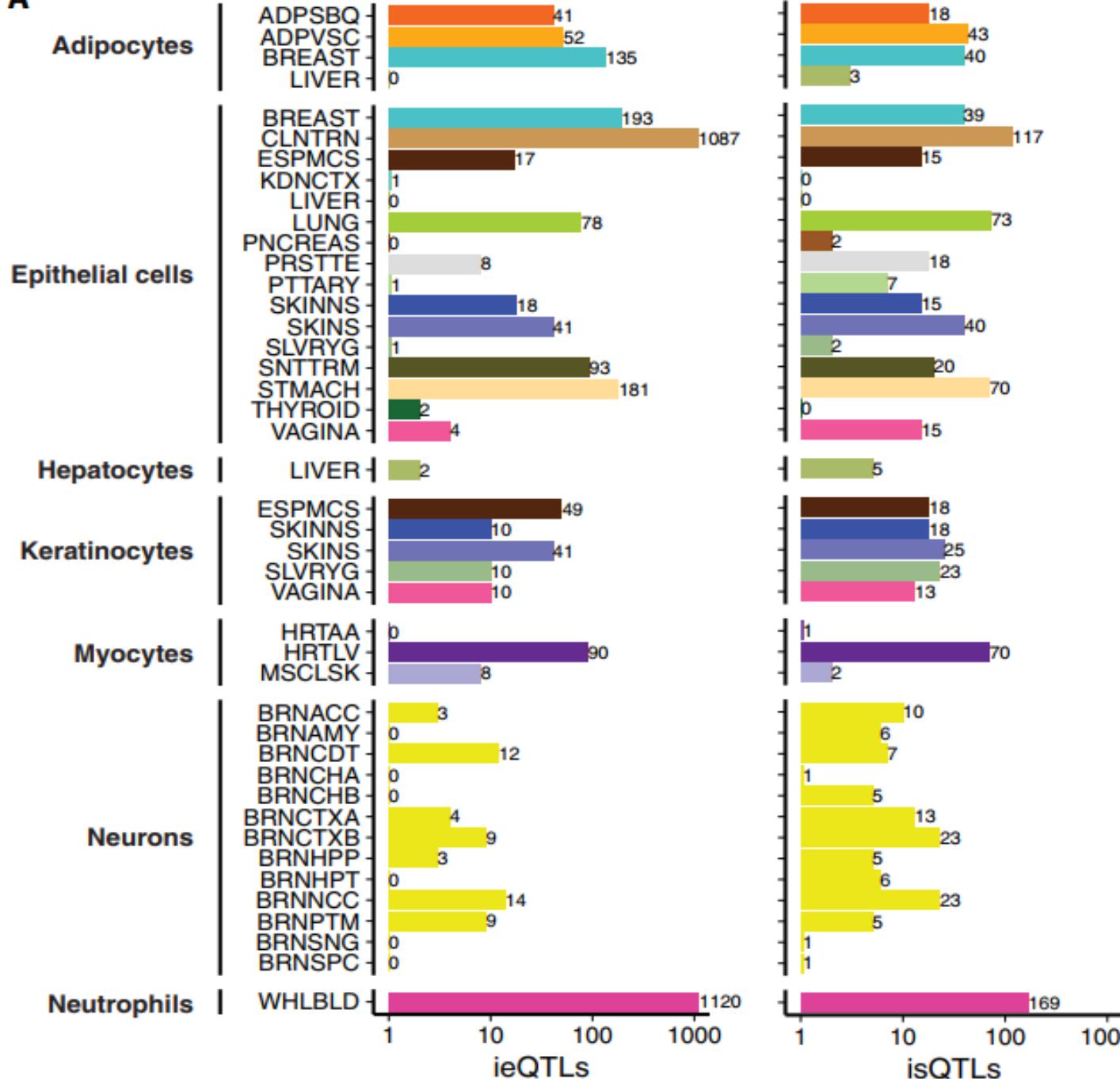
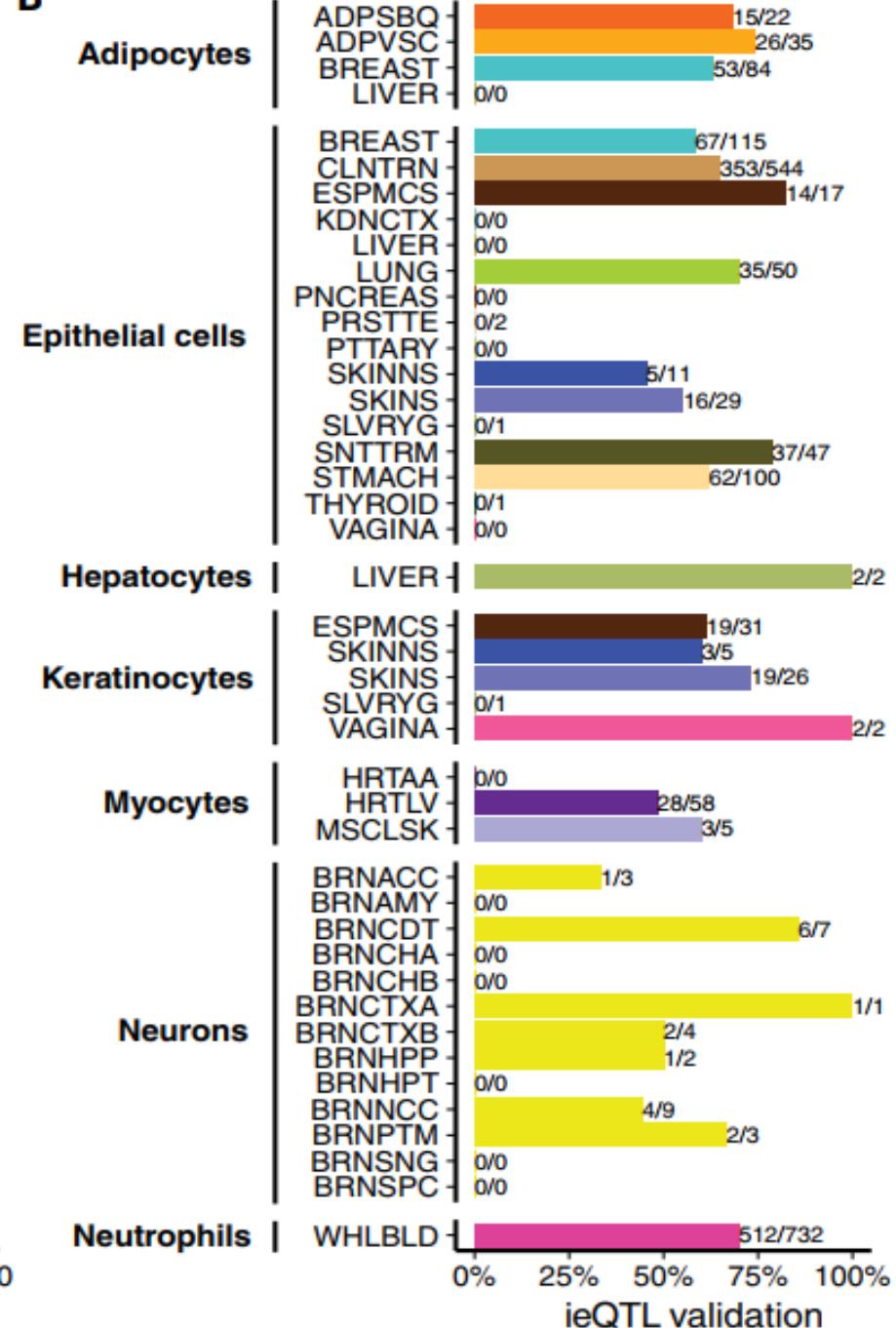
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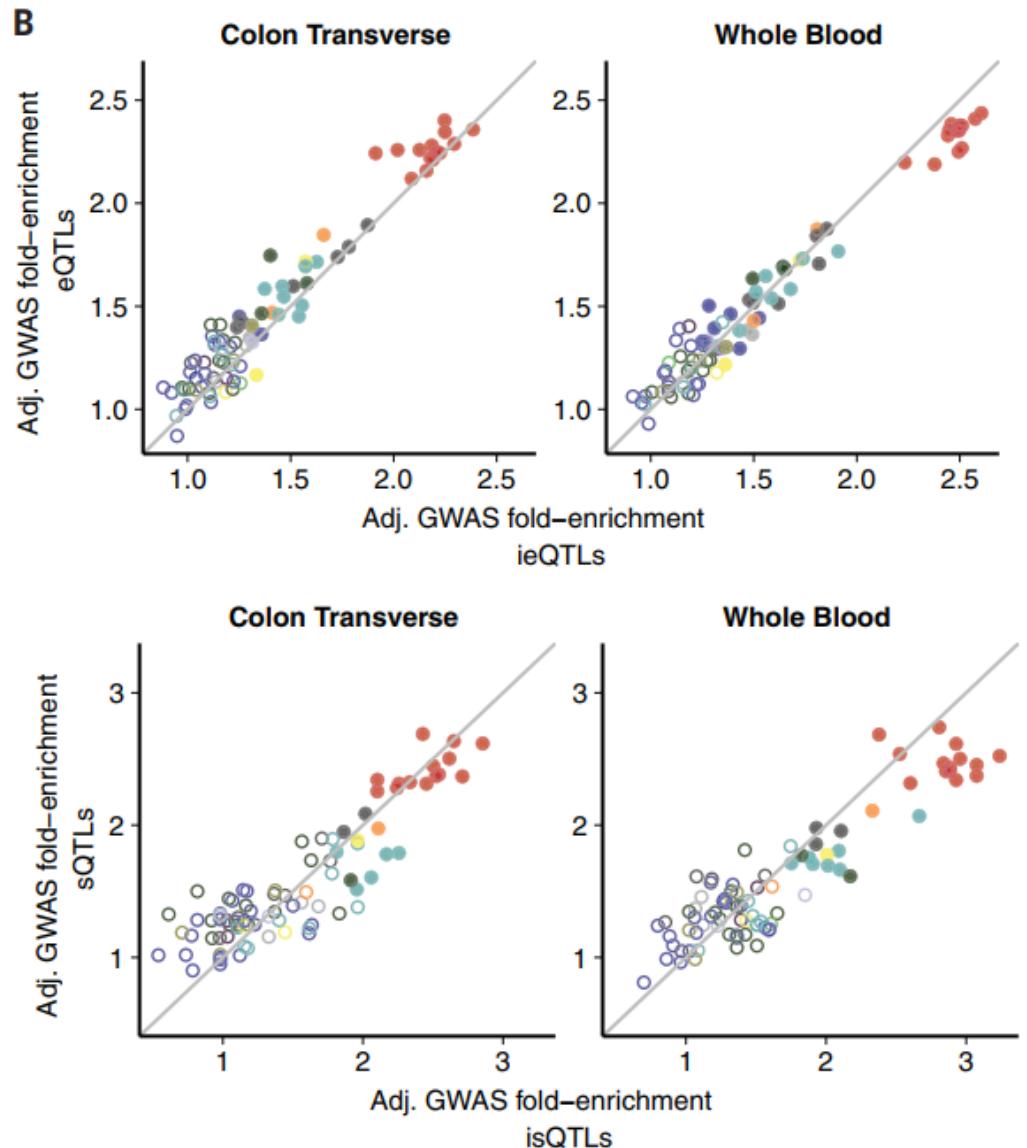
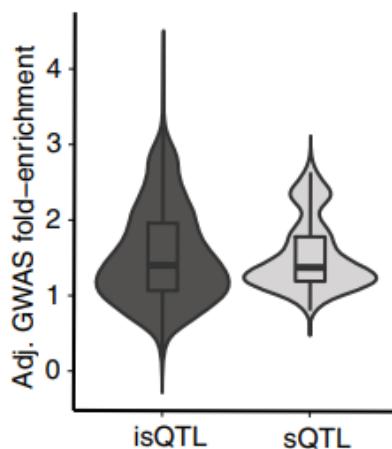
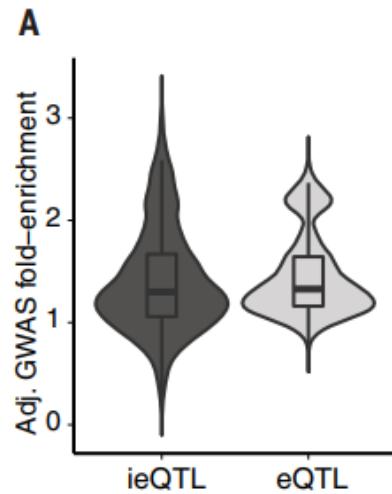


A

- 11 Brain regions
- Pituitary
- Thyroid
- Lung
- Pancreas
- Kidney cortex
- Stomach
- Transverse colon
- Small intestine terminal ileum
- Prostate
- Minor salivary gland
- Esophagus mucosa
- Vagina
- Not sun-exposed skin (suprapubic)
- Sun-exposed skin (lower leg)
- Liver
- Breast mammary tissue
- Visceral omentum
- Subcutaneous adipose
- Atrial appendage
- Left Ventricle
- Skeletal muscle
- Whole blood

**B****C**

A**B**



Thanks

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