GTEx

Genetic effects on gene expression across human tissues

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报告人：冯佳武

- The Genotype-Tissue Expression (GTEx) project
- 表达数量性状基因座（expression Quantitative Trait Loci,eQTL）指的是染色体上一些能特定调控mRNA和蛋白质表达水平的区域，其mRNA/蛋白质的表达水平量与数量性状成比例关系。
- Cis-eQTL就是某个基因的eQTL定位到该基因所在的基因组区域，表明可能是该基因本身的差别引起的mRNA水平变化
- Trans-eQTL是指某个基因的eQTL定位到其他基因组区域，表明其他基因的差别控制该基因mRNA水平的差异。
- LincRNA即Long intergenic non-coding RNA

- **The impact of rare variation on gene expression across tissues**
- **Dynamic landscape and regulation of RNA editing in mammals**
- **Landscape of X chromosome inactivation across human tissues**
- **The impact of structural variation on human gene expression**

Expression QTLs across human tissues

Allele-specific expression across human tissues

Tissue-sharing and specificity of eQTLs

Functional characterization of cis-eQTLs

Functional characterization of trans-eQTLs

Replication of eQTLs

Expression QTLs and complex disease associations

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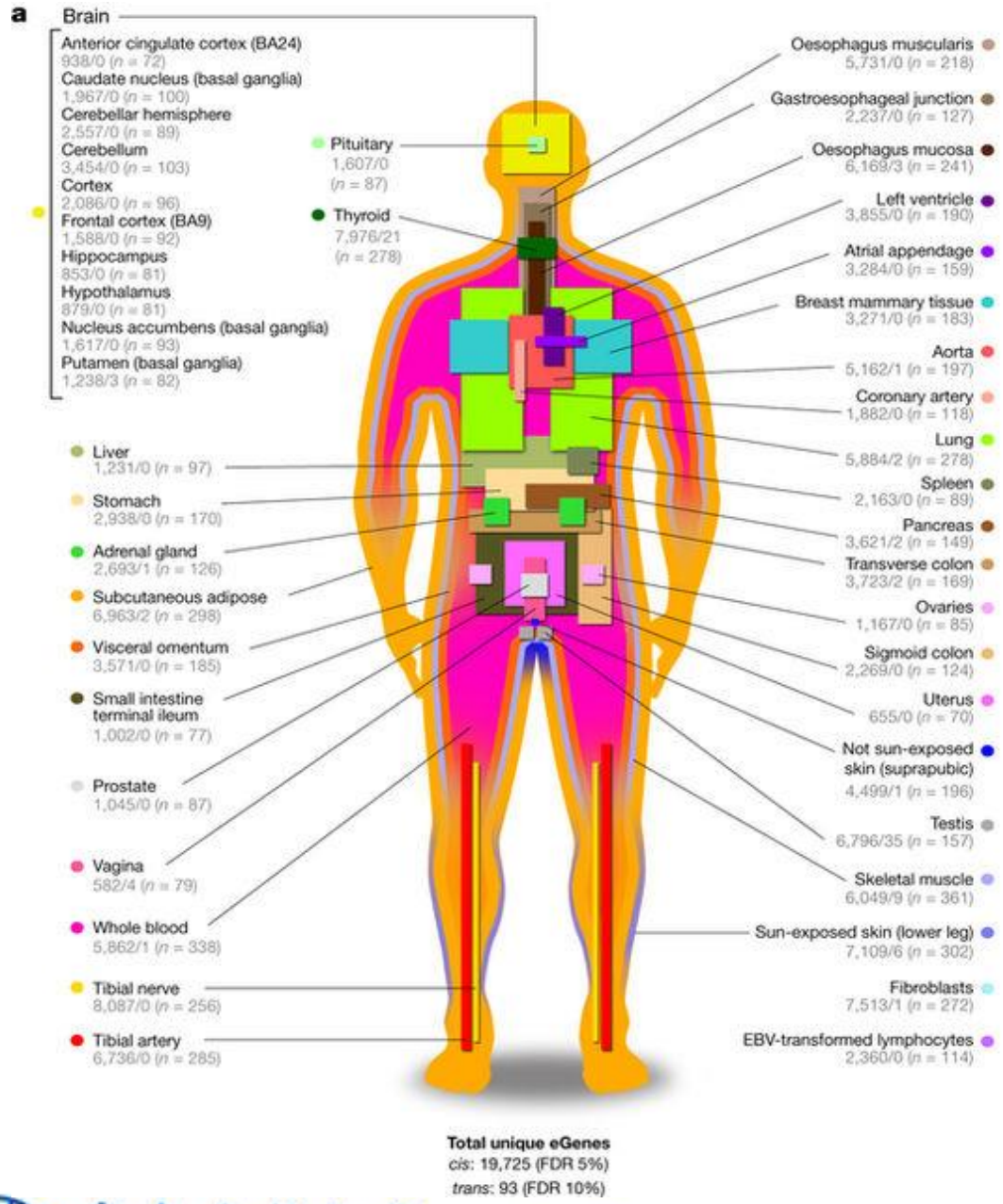

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Using probabilistic estimation of expression residuals (PEER) to obtain increased power and interpretability of gene expression analyses

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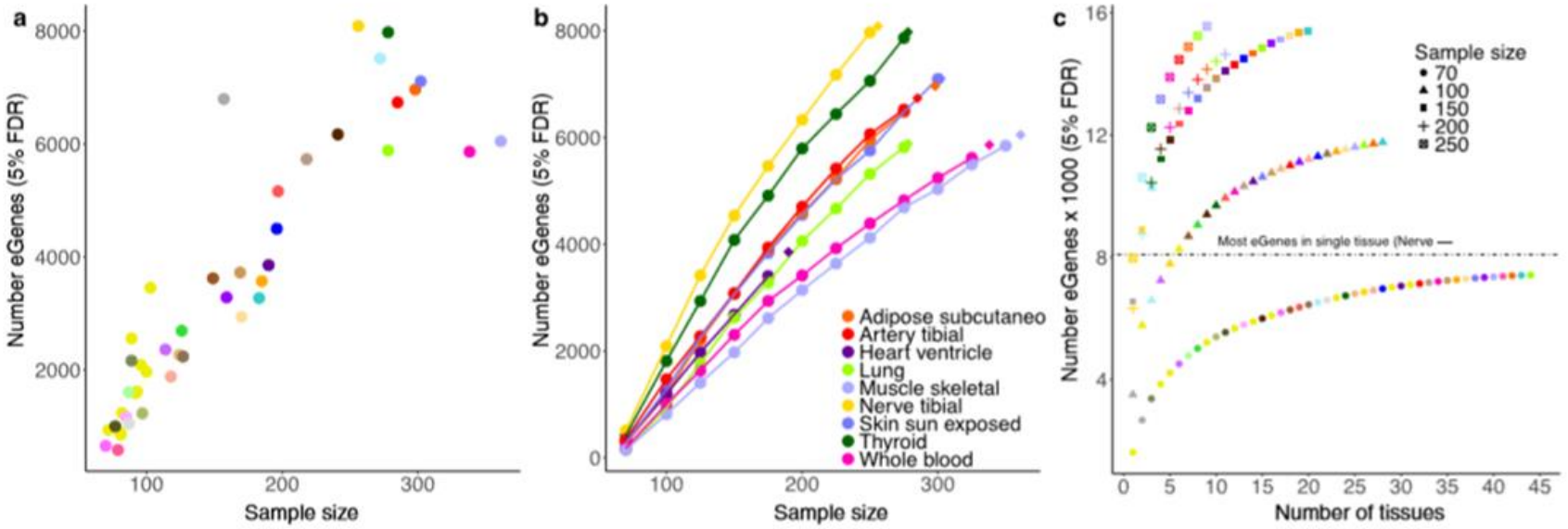

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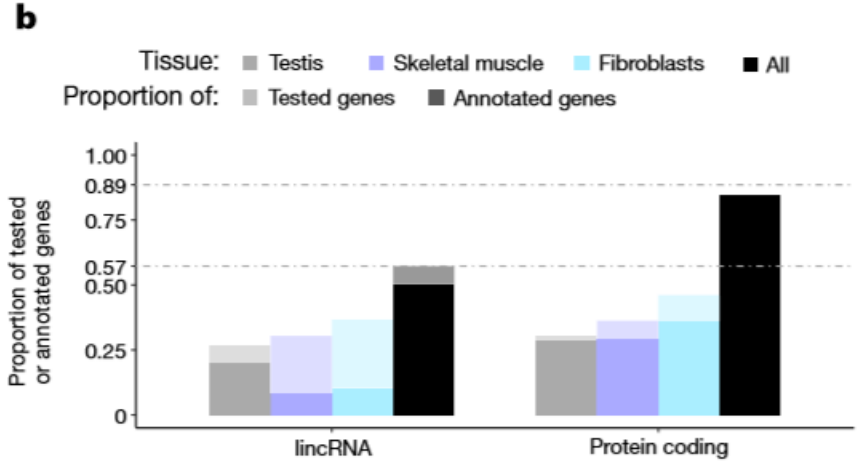

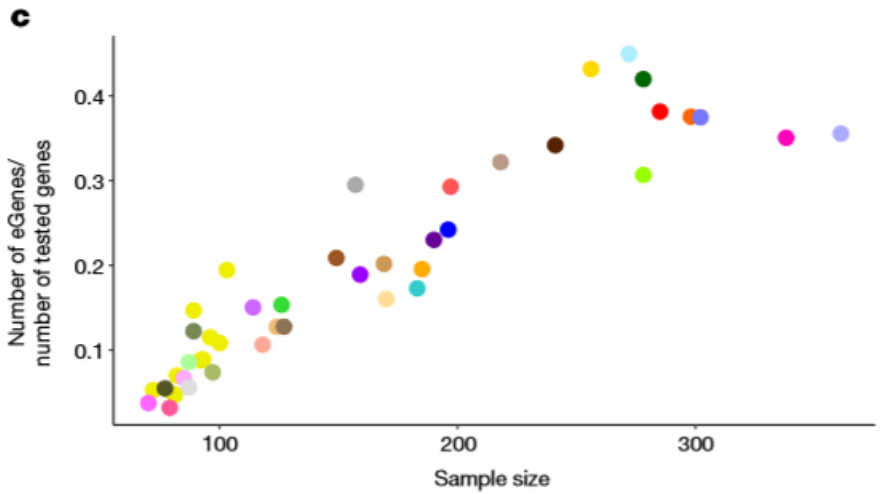

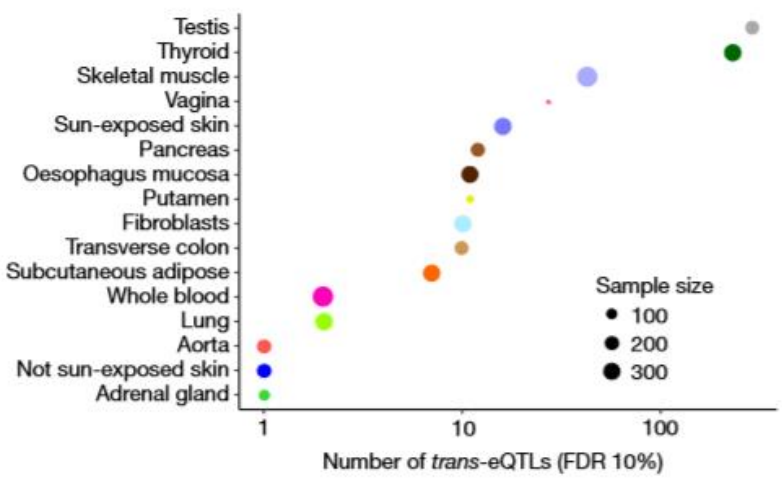

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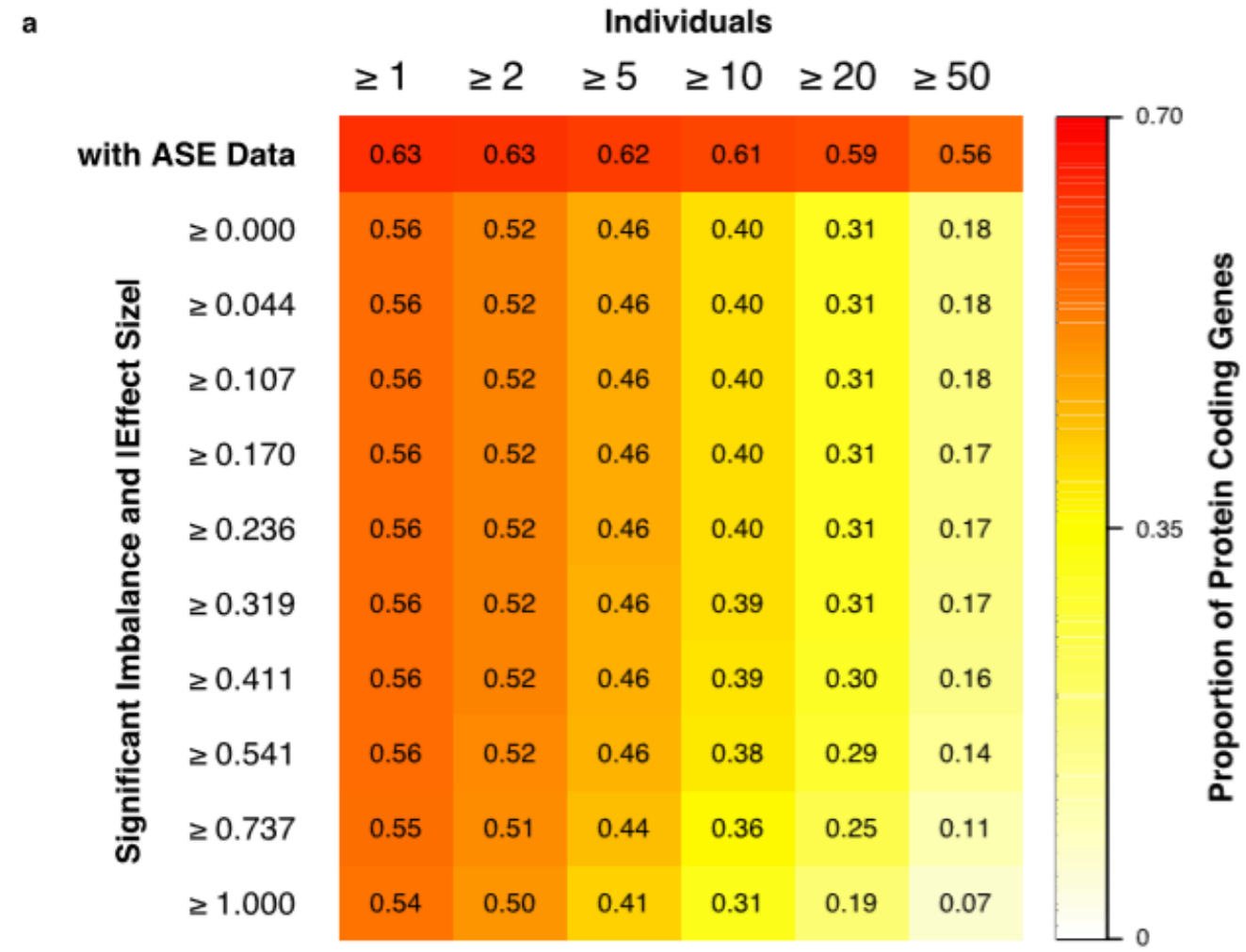

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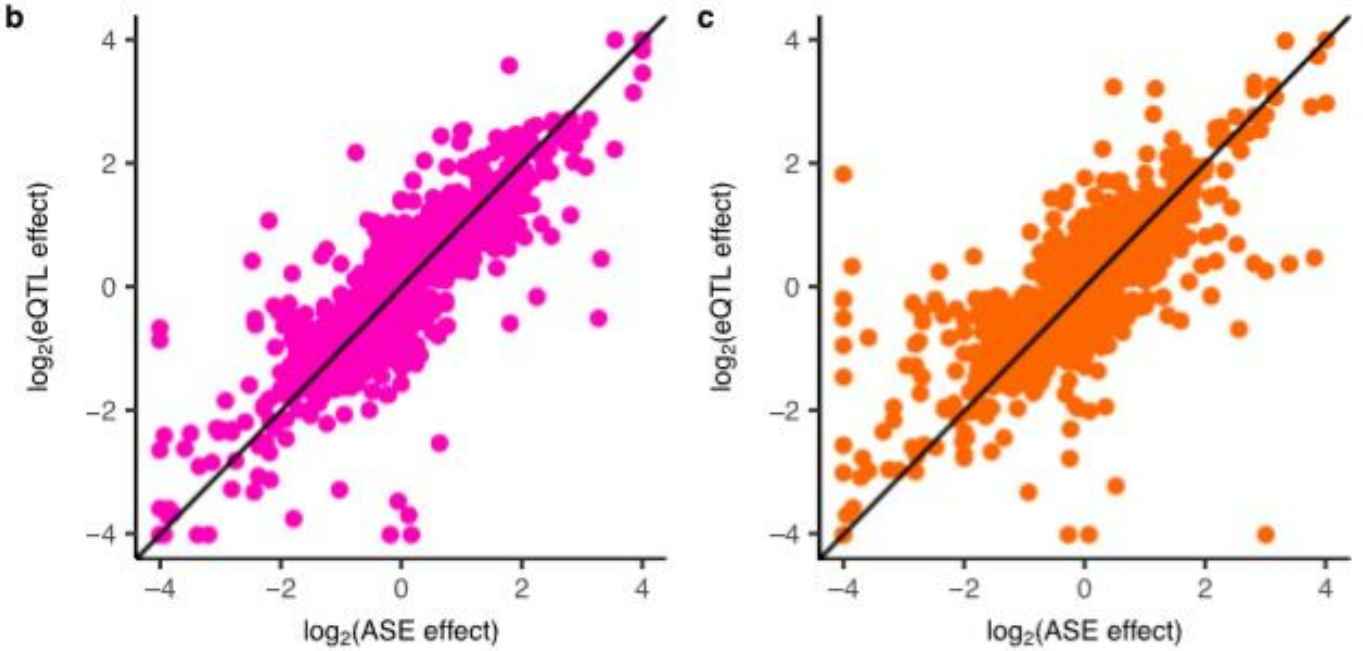

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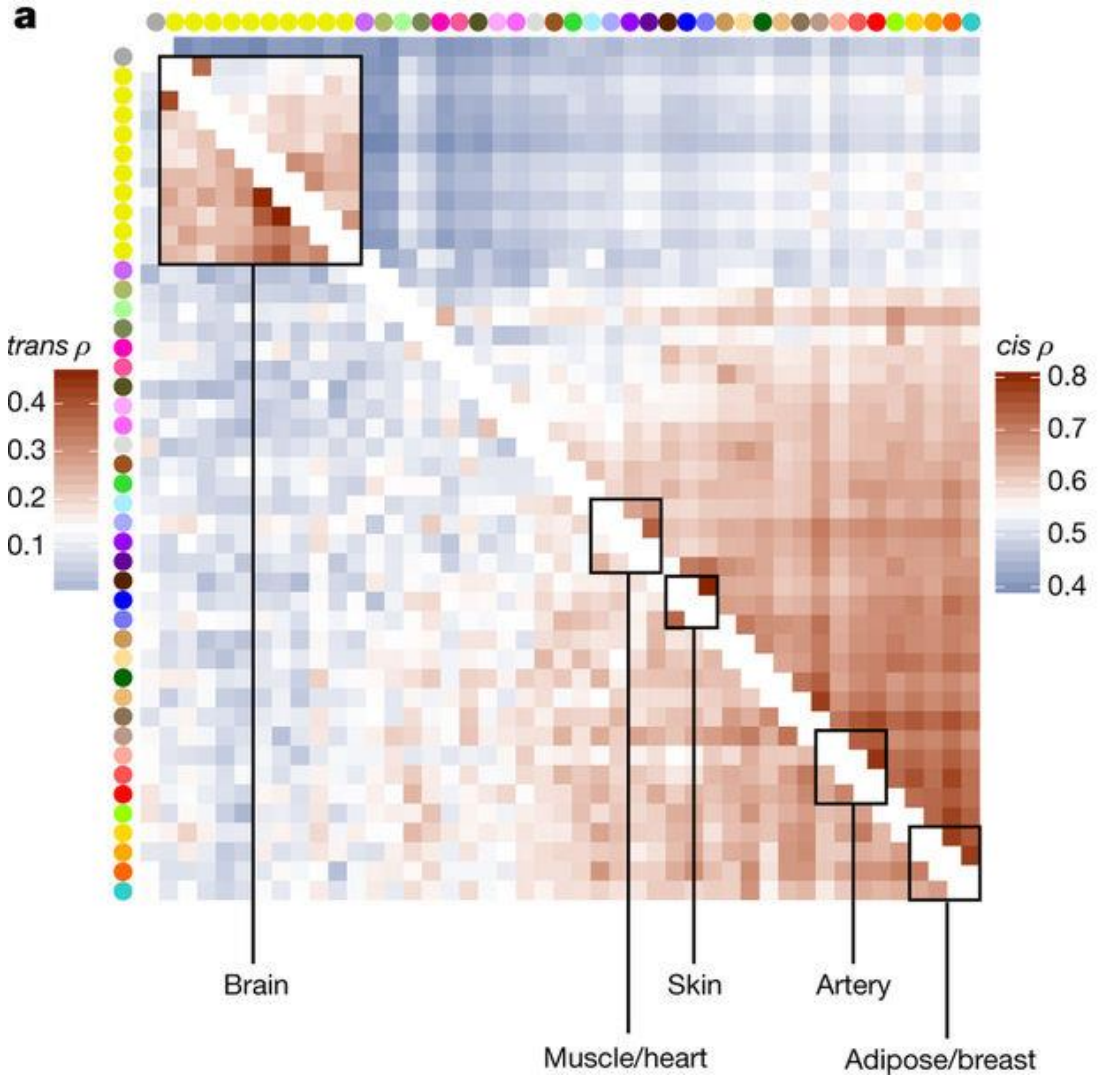

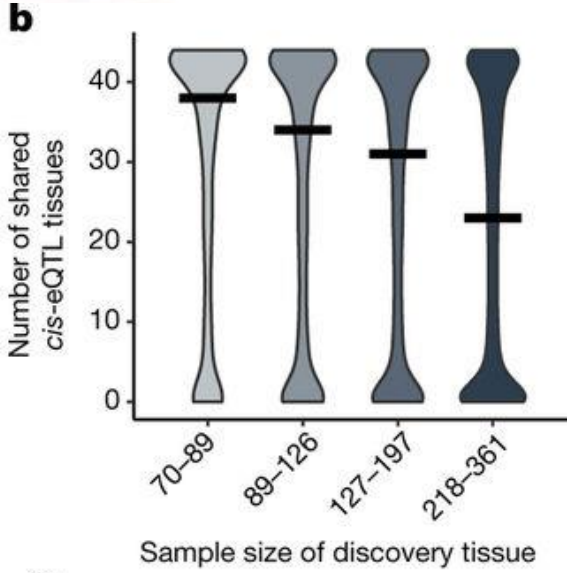

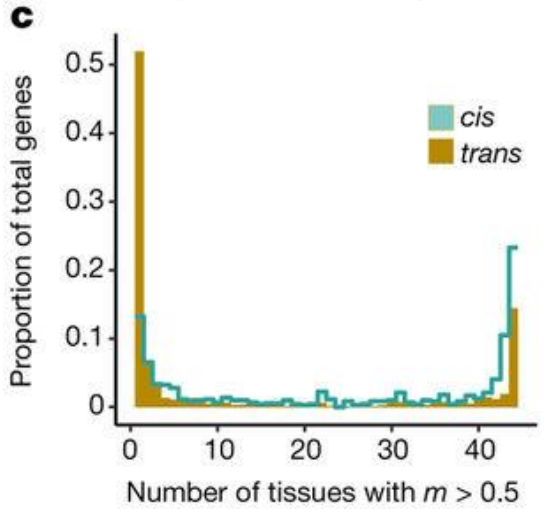

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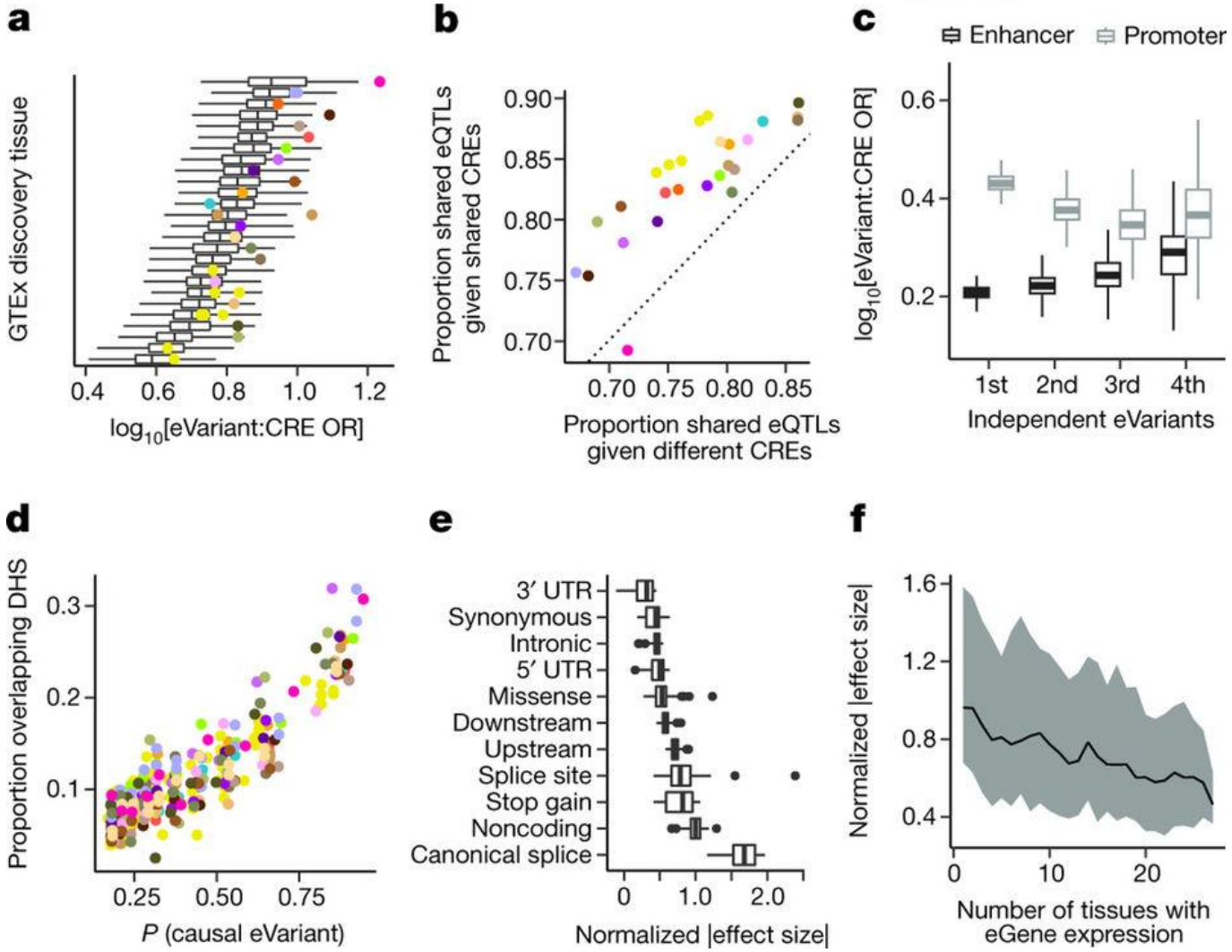

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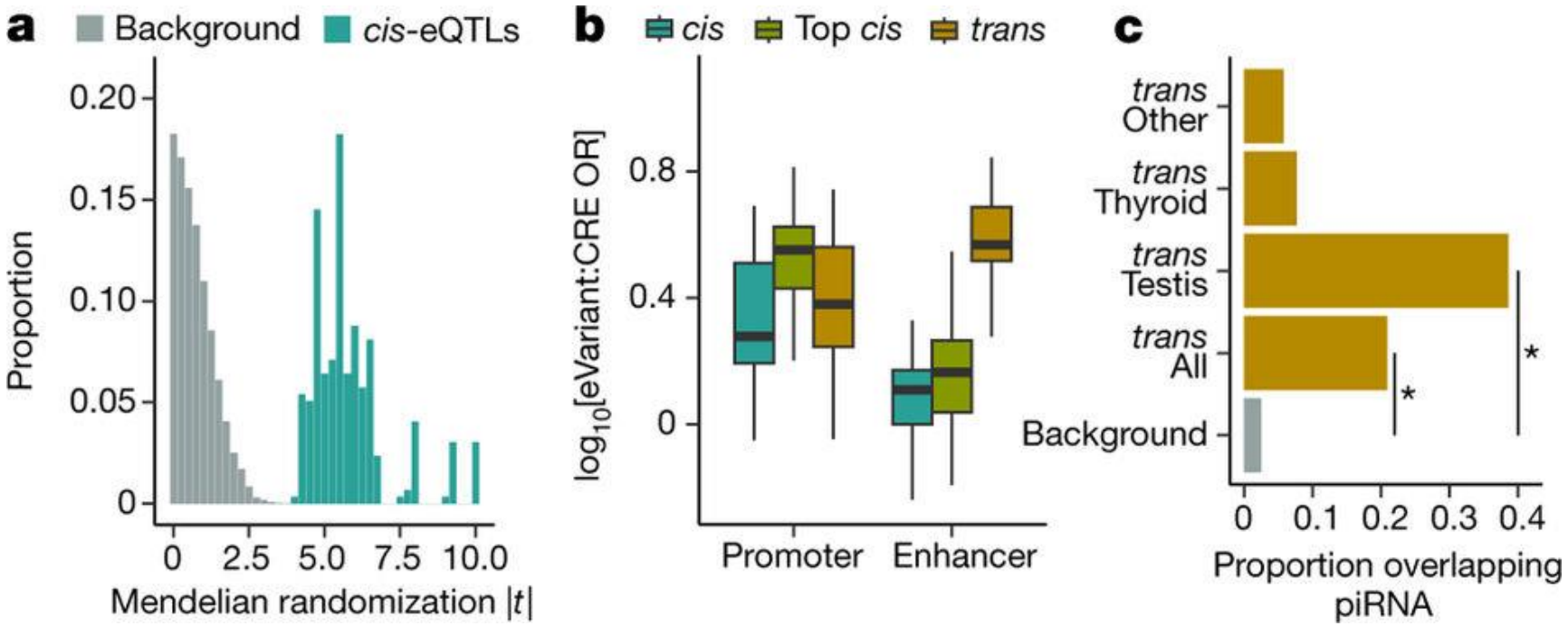

Piwi-interacting RNAs (piRNAs)

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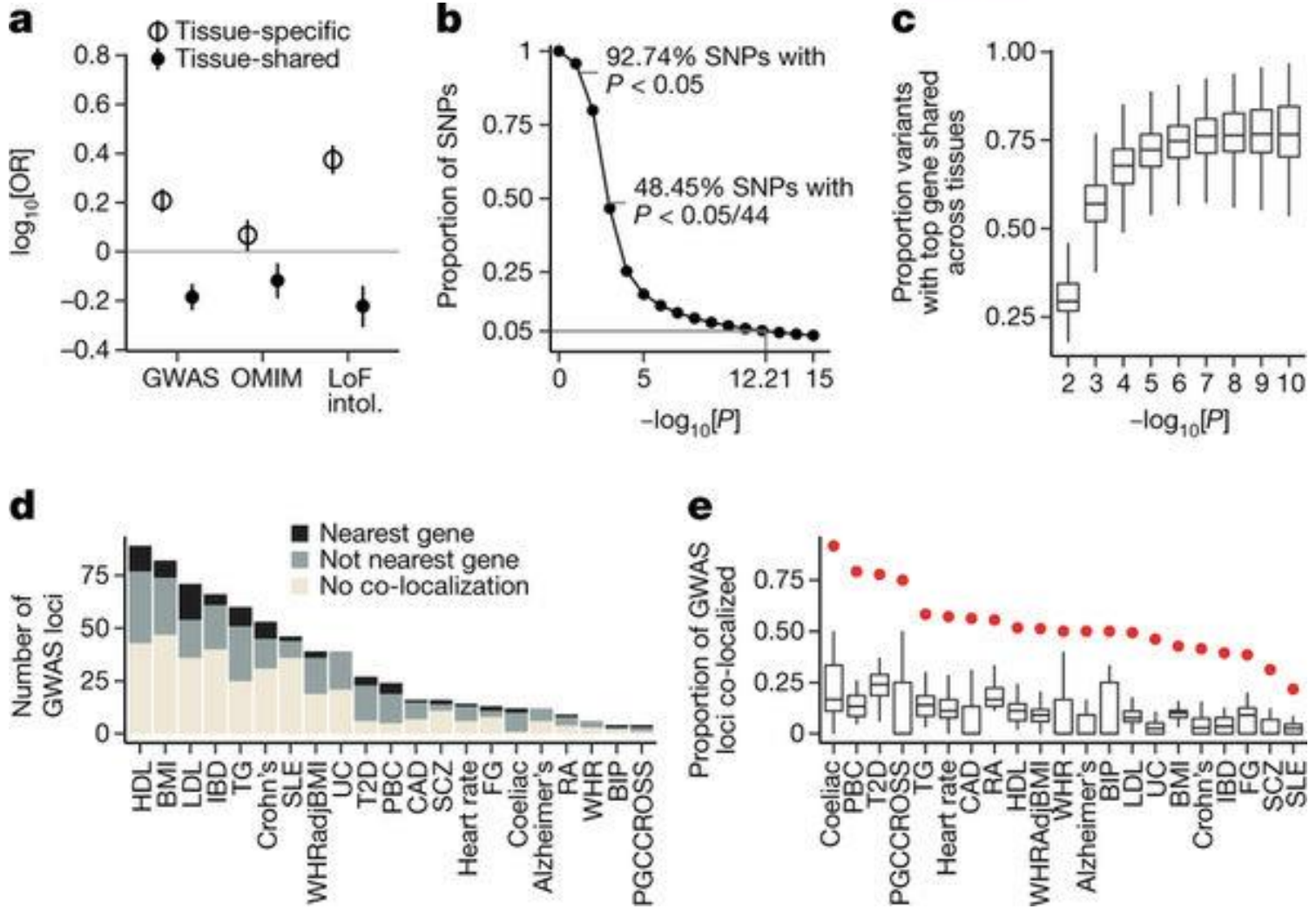

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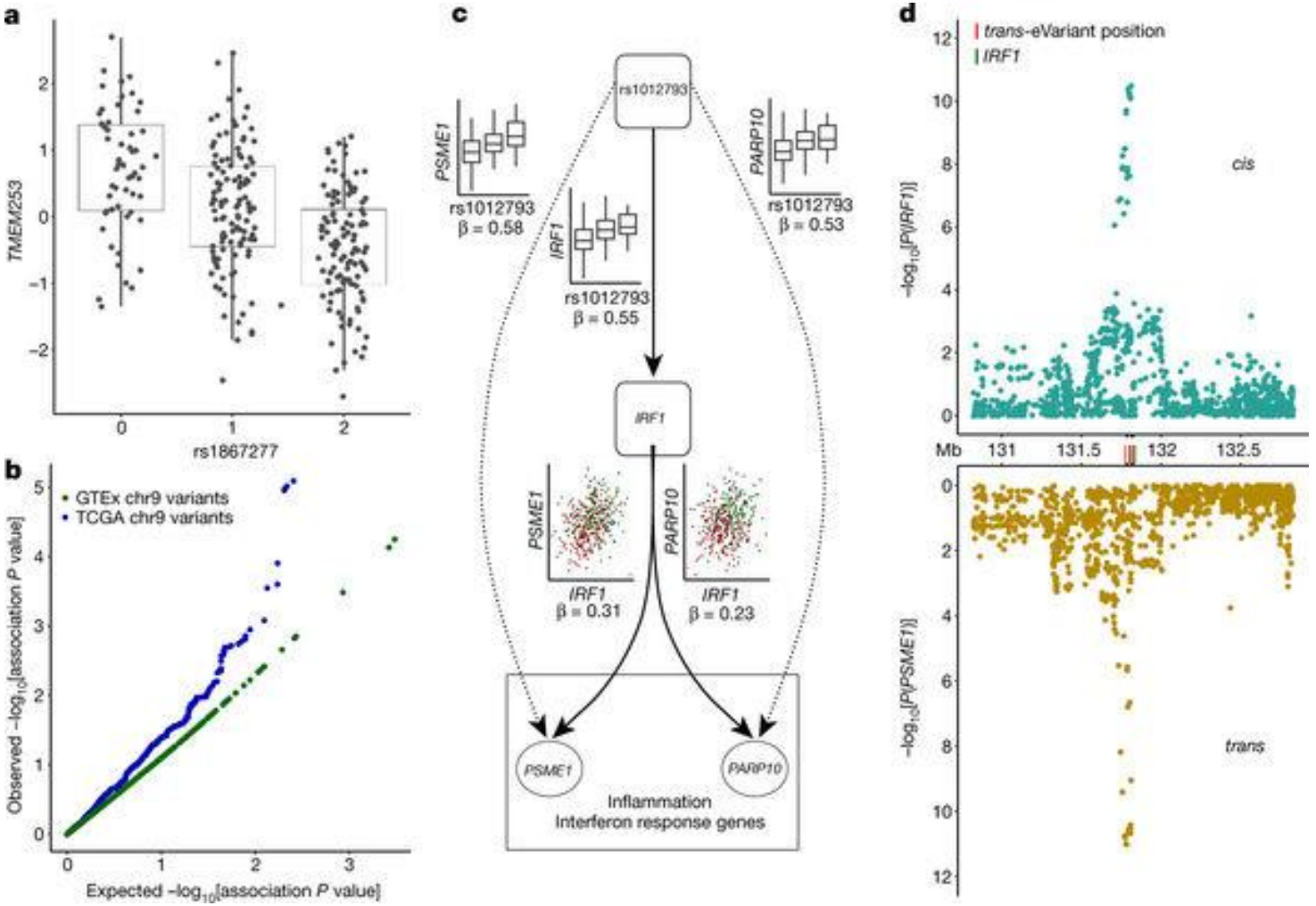

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三个问题回答

- 大规模RNA-seq数据分析了在人体组织中的基因表达的遗传效应（主要是针对e-QTL对基因表达的影响，顺式作用的遗传变异倾向于影响大多数组织或少数组织。相反，鉴定的trans-eQTL效应往往是组织特异性的，相应地在增强子区域显示出更大的富集。）
- 实验设计的严谨性和可扩展型，对统计方法的创新，对植物的大规模分析可能会容易一些（对于那些trans-eQTL的分析），但是投入也十分的庞大。
- 文章一些概念没有清楚的界定，对Trans-eQTL的复制问题还是需要一个更好的实验设计的。

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THANK YOU

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