

Supplementary Information: Appendix for Exploring regulation in tissues with eQTL networks.

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SI Datasets

Dataset S1: Odds ratio and p-values measuring enrichment in each chromatin state for *trans*-eQTLs in each tissue. cf. transeQTLs.epigenomic_roadmap_enrichment.csv file.

Dataset S2: Proportion of SNPs that are significantly associated to at least one gene depending on FDR. cf. proportion_eqtls.csv

Dataset S3: GO biological processes enrichment for each community in each tissue. cf. GeneOntology_biological_processes.csv file.

Dataset S4: Association with traits and diseases of SNPs and genes from heart left ventricle community 86 and involved in cellular respiration biological processes. cf. list_snps_genes_heart_left_ventricle_86.csv file.

Dataset S5: Enrichment in epigenomics roadmap chromatin states for central SNPs. cf. centrality_epigenomic_roadmap_enrichment.csv file.

SI Figures

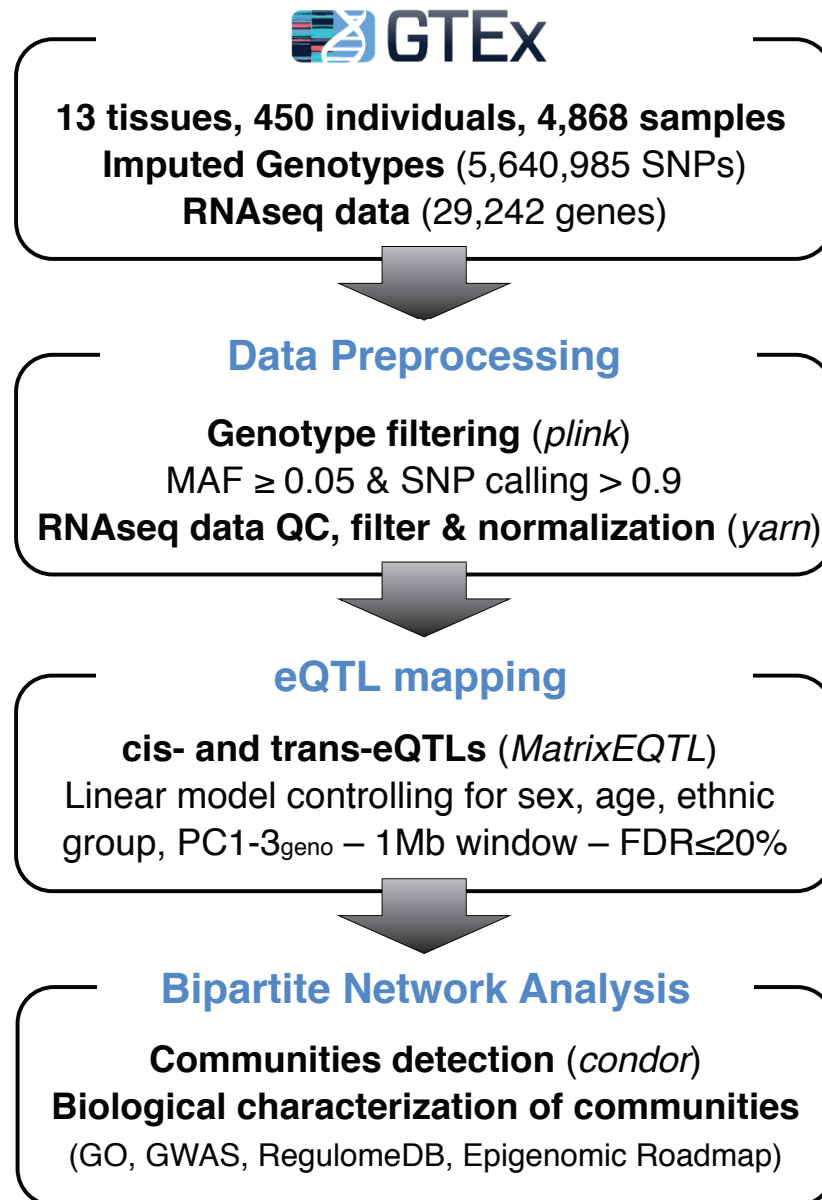


Figure S1: Study overview. Principal components 1-3 used as covariates in eQTL mapping are presented in Figure S3. Comparison of our eQTL mapping results with those from the GTEx consortium are presented in Table S2.

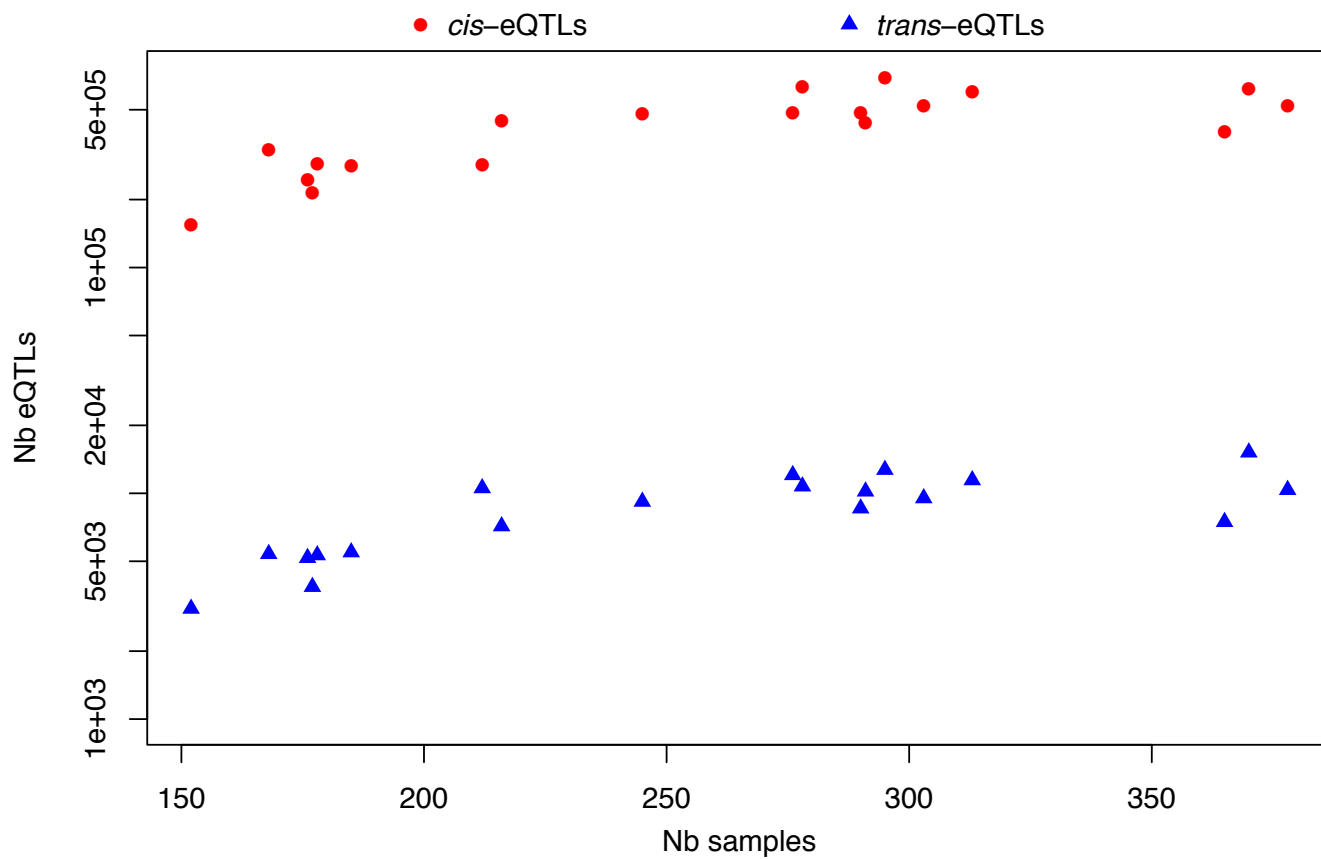


Figure S2: Number of *cis*- and *trans*-eQTLs in function of sample size.

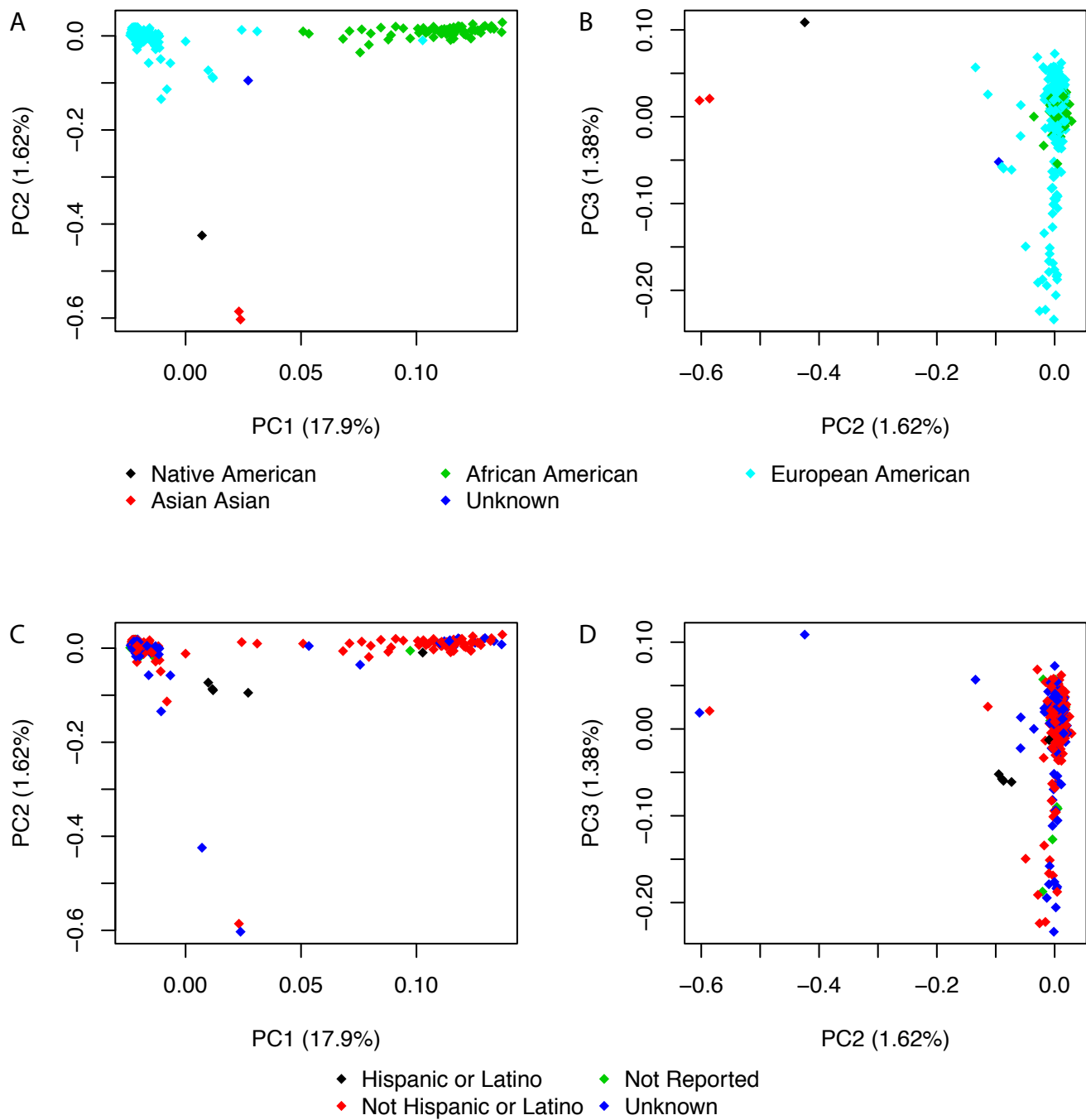


Figure S3: Principal component analysis of genotyping data. **A.** and **C.** PC1 vs. PC2. **B.** and **D.** PC2 vs. PC3. **A.** and **B.** Colors represent population. **C.** and **D.** Colors represent ethnicity as reported by donor, family/next of kin, or medical record abstraction.

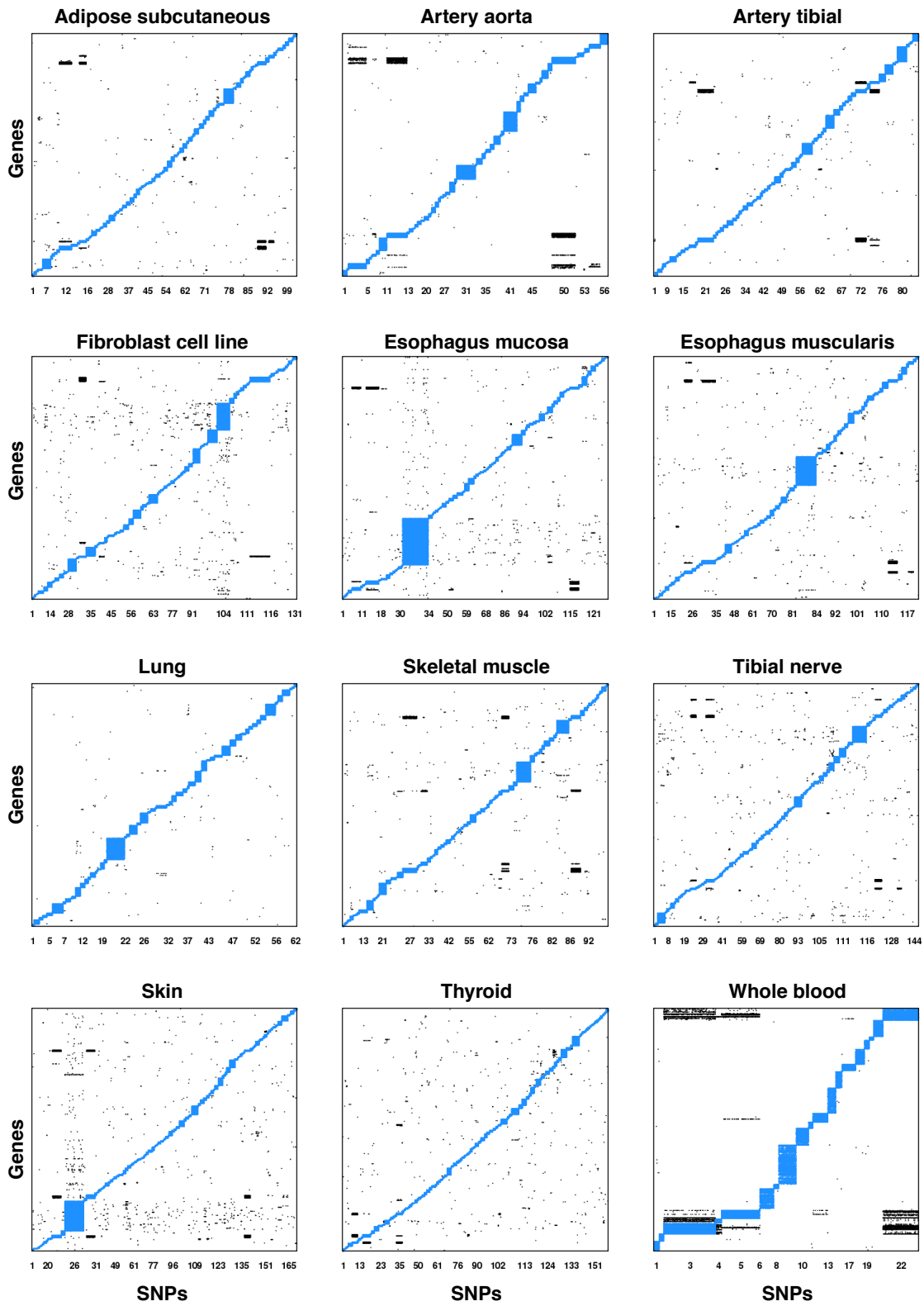


Figure S4: Structure of communities within each eQTL network. Each edge is represented by a point. Intra-community edges are plotted in blue, inter-community edges in black.

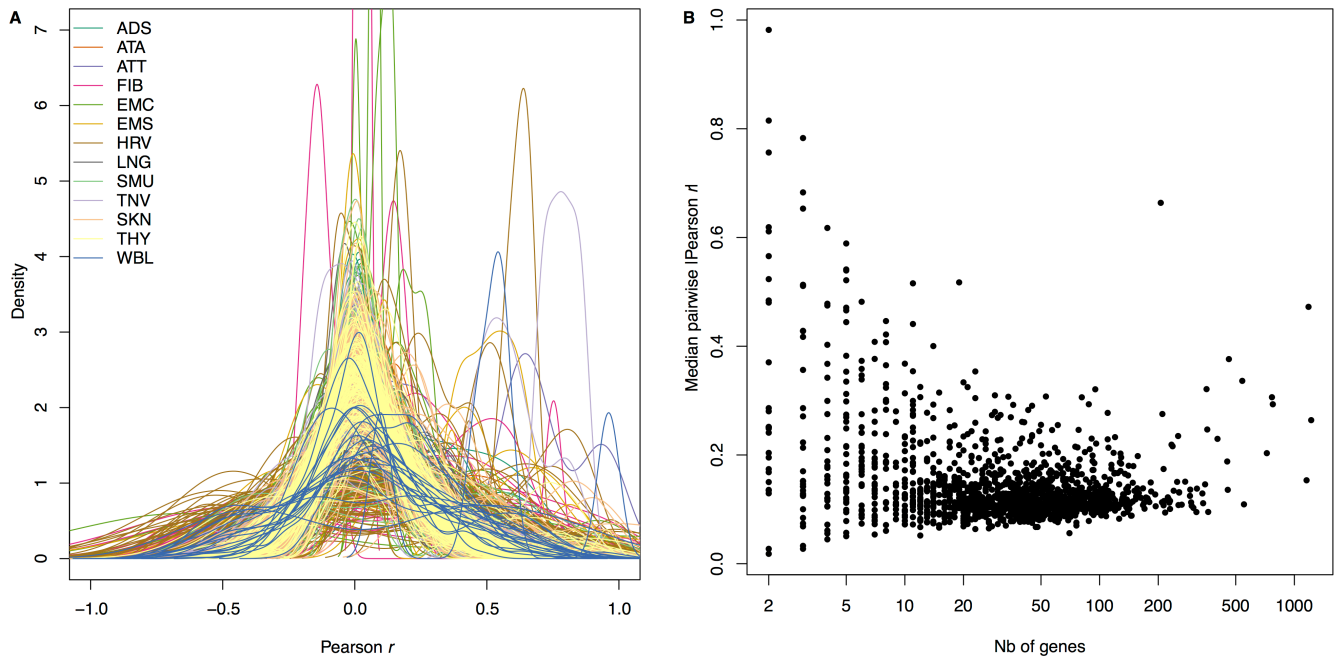


Figure S5: Gene expression correlation within communities. **A.** Distribution of pairwise Pearson correlation coefficients between gene expression levels for each community and each tissue. **B.** Distribution of median of $|\text{Pearson } r|$ in function of the community size, expressed in number of genes.

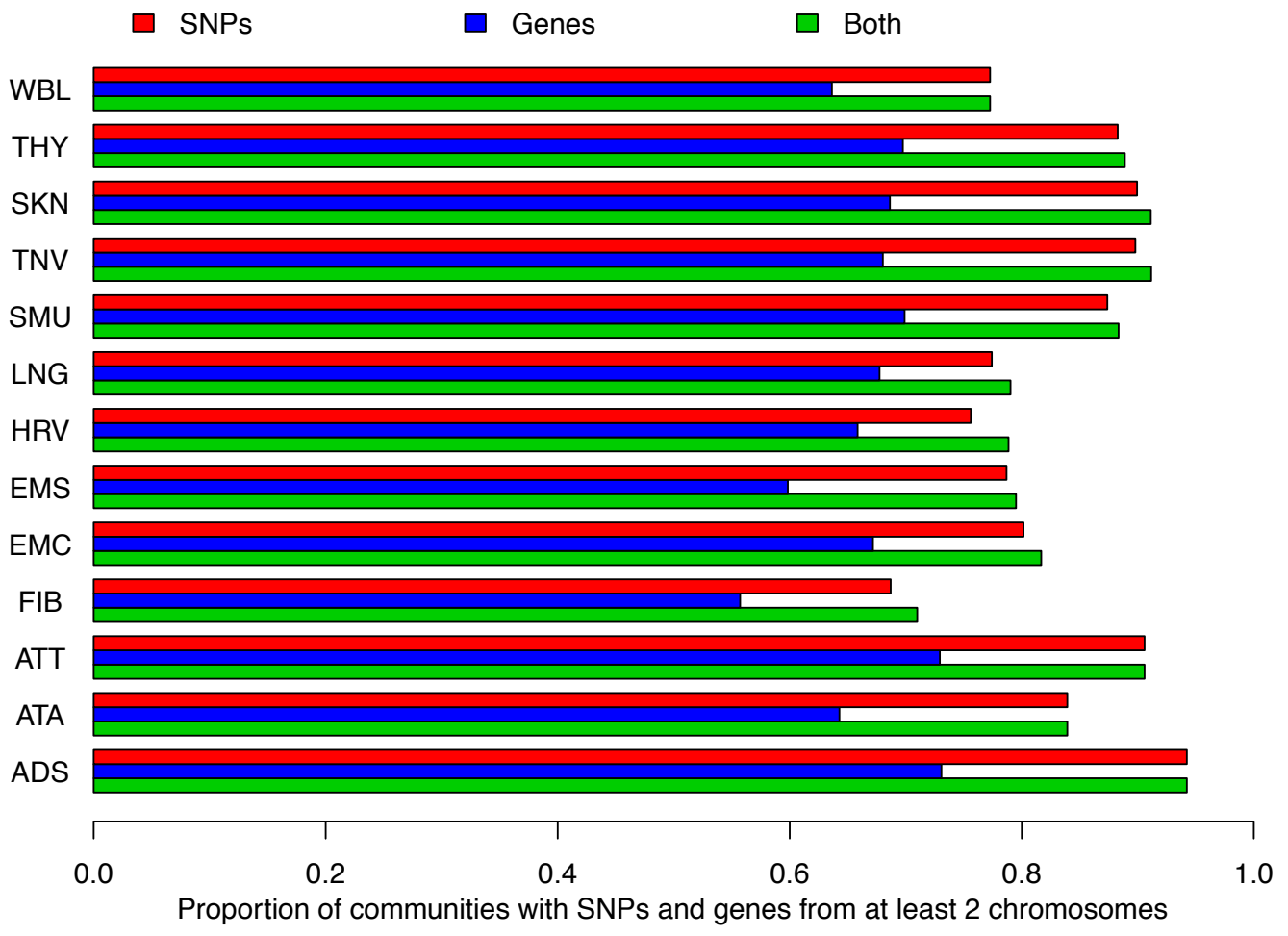


Figure S6: Proportion of communities with SNPs, genes or both mapping to at least two chromosomes.

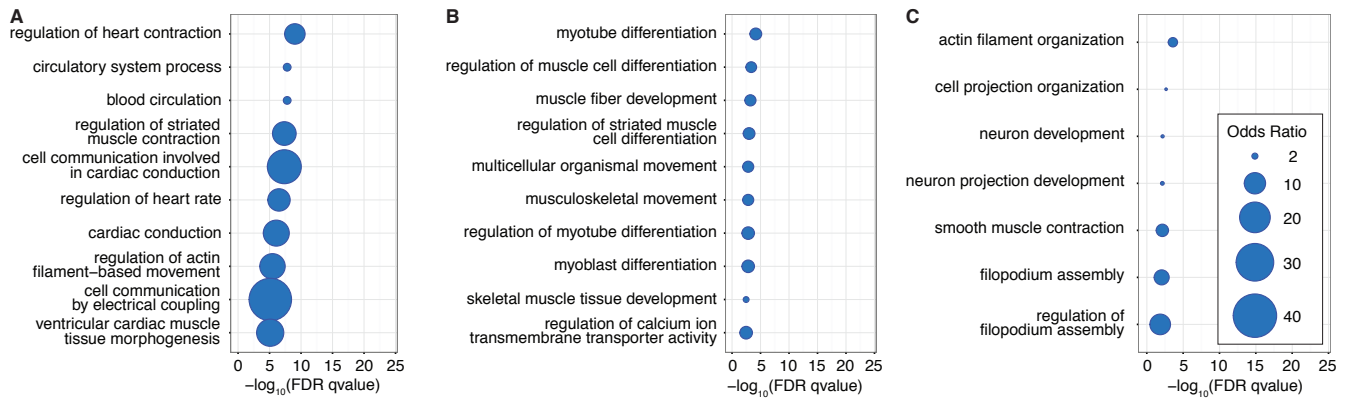


Figure S7: Enrichment in muscle-specific gene ontology terms of 3 communities. **A.** Heart left ventricle community 30 is enriched for genes involved in cardiac muscle development and contraction. **B.** Muscle skeletal community 75 is enriched for genes involved in striated muscle development. **C.** Esophagus muscularis community 83 is enriched for genes involved in smooth muscle contraction.

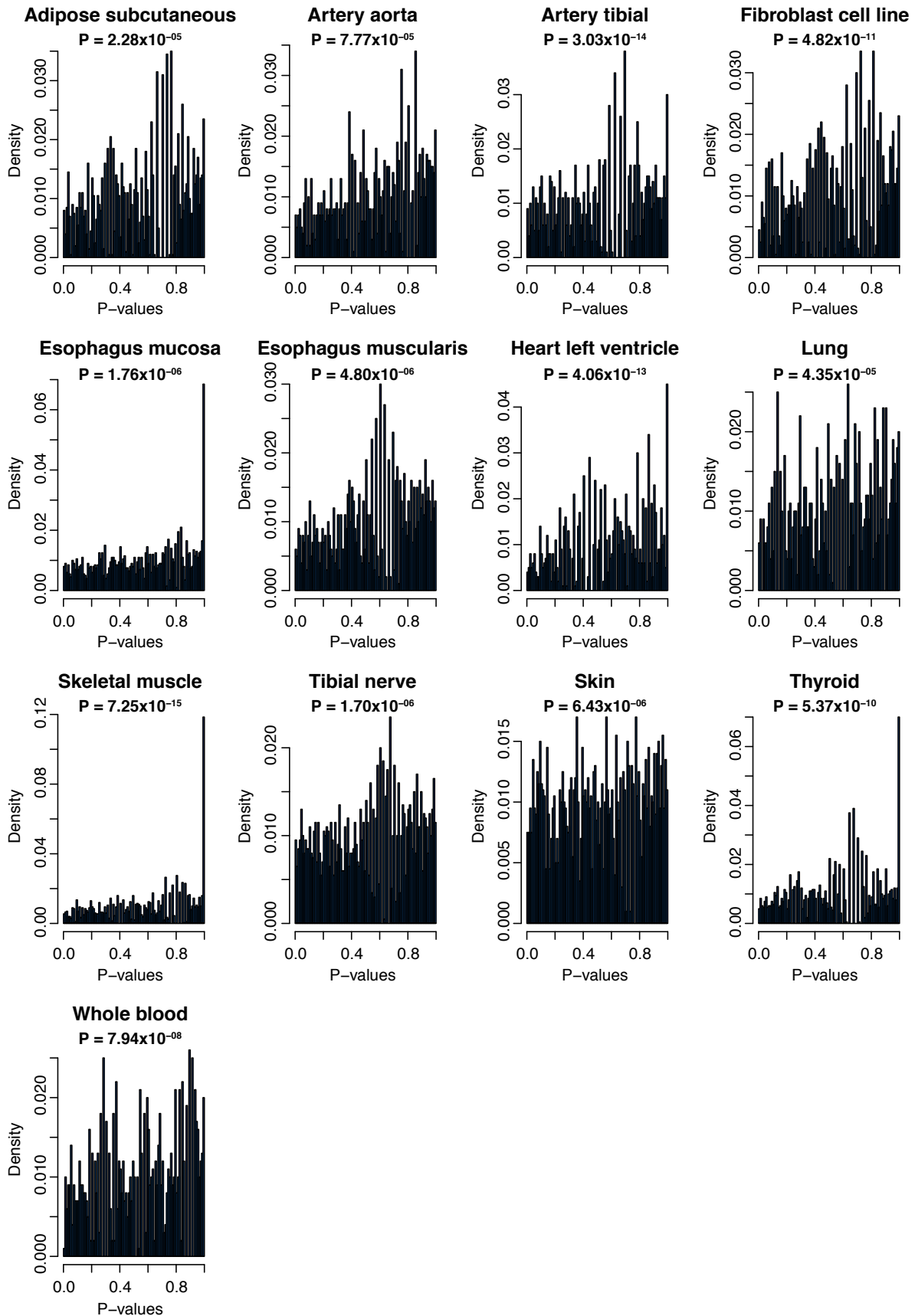


Figure S8: Null distribution of p-values for shared GO Term GO:0010468 – regulation of gene expression – obtained using 1000 resamplings. For each tissue, true p-values are written on top of each panel.

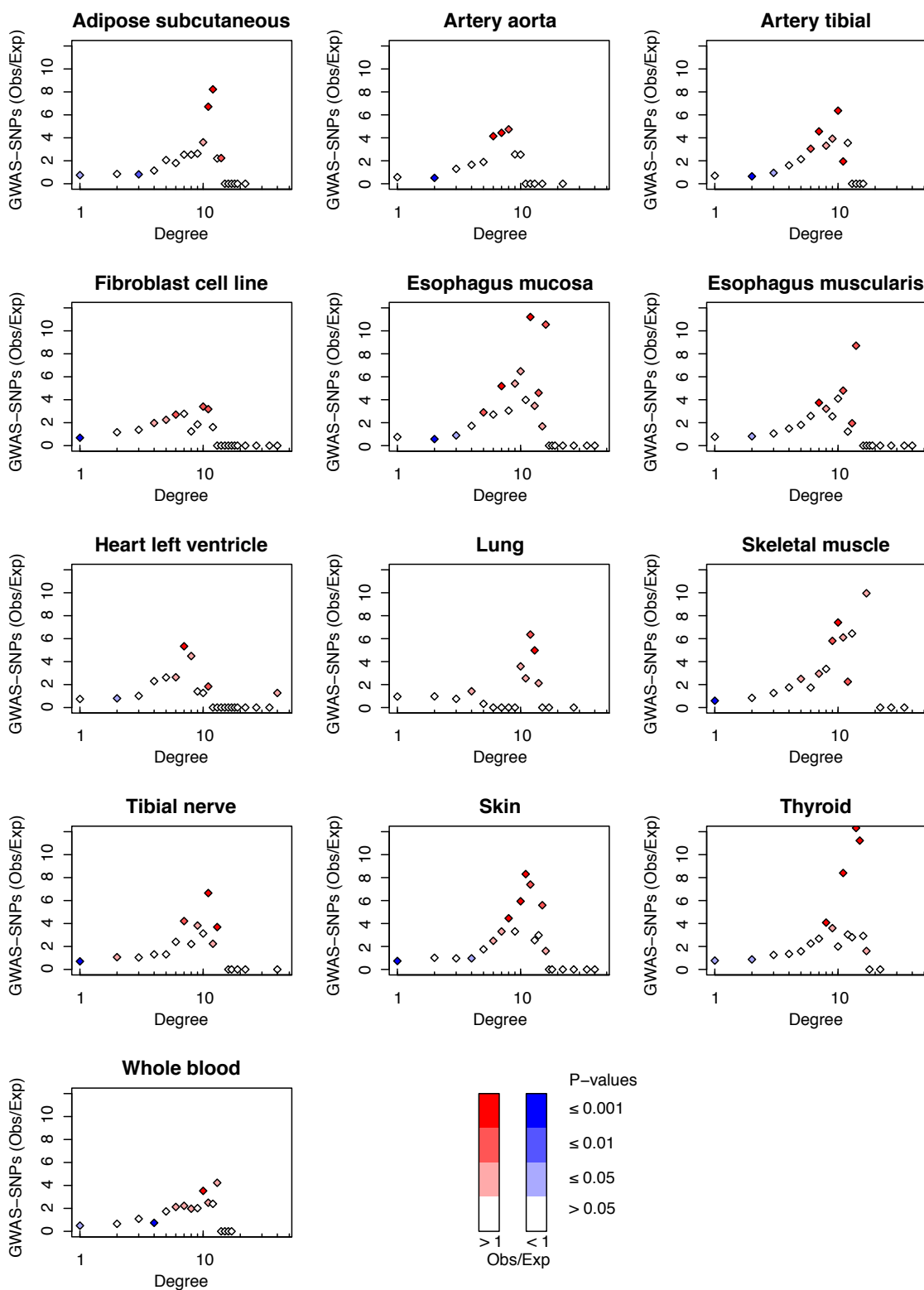


Figure S9: Ratio of observed vs. expected number of SNPs associated with all traits or diseases by GWAS studies depending on their network degree for each tissue-specific network.

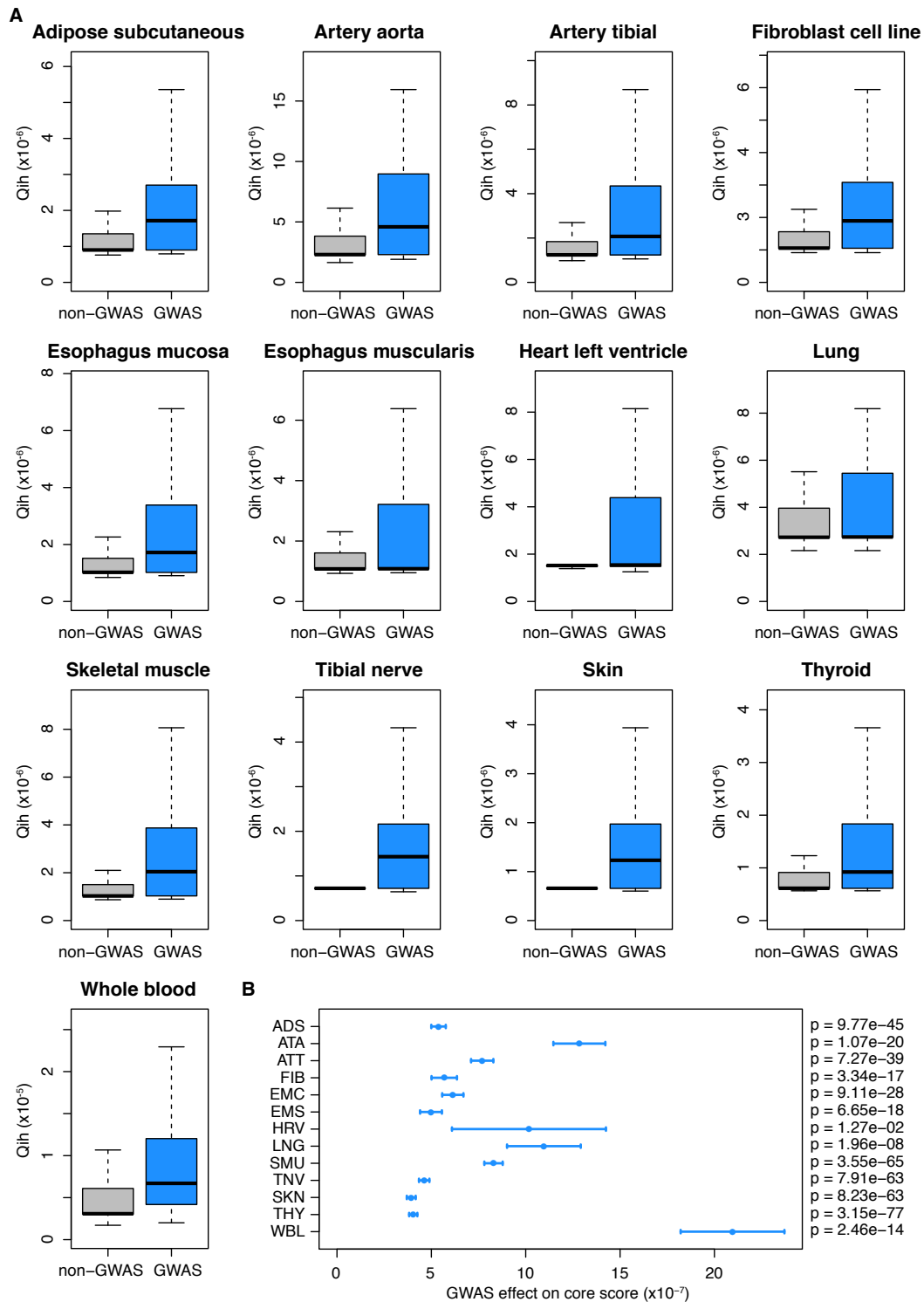


Figure S10: GWAS-SNPs are enriched in high core score. **A.** Distribution of core-scores for SNPs associated (in blue) or not (in gray) to traits or diseases by GWAS studies for all tissues. **B.** Coefficient for the GWAS term in the linear regression model (Eq. 4). Bars indicate the standard error for the coefficient. *P*-values indicated on the right were obtained using the likelihood ratio test described in the Methods.

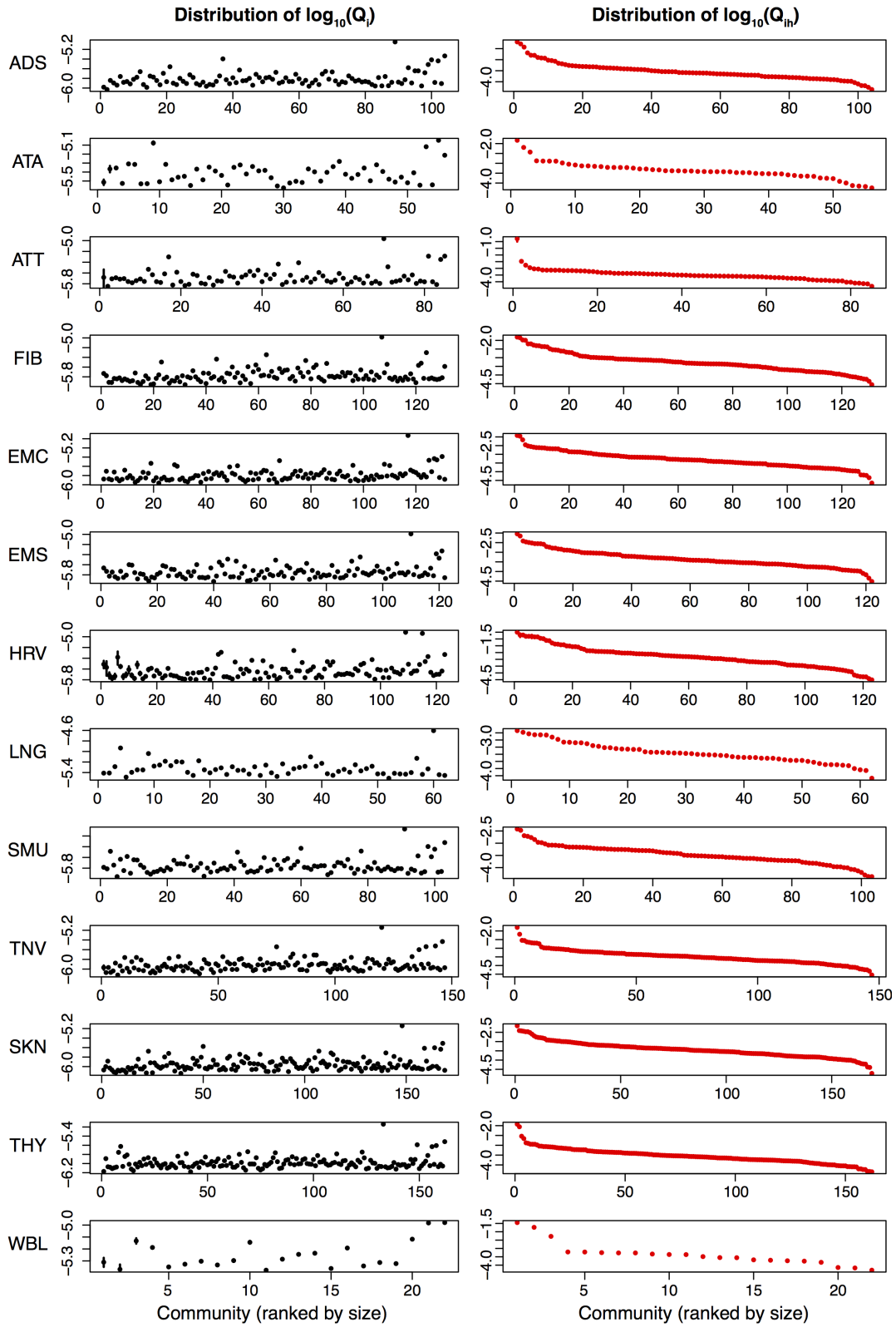


Figure S11: Distribution of Q_i and Q_{ih} in function of community size. Points represent the mean and bars indicates standard errors for each community.

SI Tables

Table S1: Summary of data: tissues and number of samples.

Tissue names	Abbreviation	GTEX tissues	Samples
Adipose subcutaneous	ADS	Adipose - Subcutaneous	313
Aorta	ATA	Artery - Aorta	216
Artery tibial	ATT	Artery - Tibial	303
Fibroblast	FIB	Skin cells - Transformed fibroblasts	291
Esophagus mucosa	EMC	Esophagus - Mucosa	276
Esophagus muscularis	EMS	Esophagus - Muscularis	245
Heart left ventricle	HRV	Heart - Left ventricle	212
Lung	LNG	Lung	290
Skeletal muscle	SMU	Muscle - Skeletal	378
Tibial nerve	TNV	Nerve - Tibial	278
Skin	SKN	Skin - Not sun exposed (Suprapubic)	127
		Skin - Sun exposed (Lower leg)	243
		Total	370
Thyroid	THY	Thyroid	295
Whole blood	WBL	Whole blood	365

Table S2: Comparison of our results (at a FDR < 0.05) with GTEX *cis-eQTL* list.

Tissue names	GTEX	Our analysis	Common eQTLs	Proportion
Adipose subcutaneous	1,311,216	599,385	445,659	74.35%
Artery aorta	878,091	446,775	313,000	70.06%
Artery tibial	1,245,107	520,345	388,842	74.73%
Fibroblast cell line	1,315,975	438,548	330,338	75.33%
Esophagus mucosa	1,118,435	484,212	350,936	72.48%
Esophagus muscularis	1,019,564	481,827	339,745	70.51%
Heart left ventricle	620,472	285,283	201,318	70.57%
Lung	1,095,502	483,266	355,454	73.55%
Skeletal muscle	1,124,399	523,434	382,387	73.05%
Tibial nerve	1,491,673	630,254	466,921	74.08%
Skin	1,450,025	617,307	445,664	72.19%
Thyroid	1,592,982	691,333	525,681	76.04%
Whole blood	1,060,536	397,941	298,012	74.89%