

## Summary data on fQTLs calculated from 8 cell types

Cell_type	num_fQTLs	min_bonf_corrected_p_val	max_bonf_corrected_p_val
Ex4	630	1.46E-23	0.049764449
Ex3	126	1.93E-09	0.042337565
Astrocytes	105	1.44E-07	0.048867252
Microglia	91	4.06E-07	0.048665373
Ex5	26	1.32E-10	0.045802929
Oligodendrocytes	10	1.38E-08	0.029443349
Endothelial	8	0.021597405	0.047000607
Neuron	0	--	--

# Stats on fQTLs w/SNVs that coincide with trans-eQTLs

439/457 genes have all needed data (ENSG ID, TSS, etc)

## Results based on using 8 cell types

p_val_thresh	tot # matches	# unq matched SNVs	# unq fQTL SNVs	fract unq fQTL SNVs that match eSNVs
0.05	100100	<b>697</b>	726	<b>0.96</b>
0.1	113433	843	898	0.939
0.2	126070	996	1089	0.915
0.3	133431	1113	1242	0.896

\* P-values are Bonferroni-corrected (for both fQTLs and trans-eQTLs)

## Trans-eGenes associated with the 20 most significant fQTLs (linked through common SNV) [8 cell analysis]

ATP13A4 ATPase 13A4  
CDC14A cell division cycle 14A  
CLDN11 claudin 11  
CPNE4 copine 4  
DPYD dihydropyrimidine dehydrogenase  
GPR153 G protein-coupled receptor 153  
IL1RAP interleukin 1 receptor accessory protein  
LMO4 LIM domain only 4  
LRRC38 leucine rich repeat containing 38  
MAP6D1 MAP6 domain containing 1  
MSX1 msh homeobox 1  
PLD1 phospholipase D1  
PTPRU protein tyrosine phosphatase, receptor type U  
RGS12 regulator of G protein signaling 12  
SORCS2 sortilin related VPS10 domain containing receptor 2  
SOX2-OT SOX2 overlapping transcript  
SST somatostatin  
STK32B serine/threonine kinase 32B  
TF transferrin  
TMCO4 transmembrane and coiled-coil domains 4

# Most significant GO Processes among top 1000 fQTLs matched to trans-eQTLs [8 cell analysis]

synaptic transmission, GABAergic  
forebrain neuron fate commitment  
axonal fasciculation  
neuron projection fasciculation  
regulation of neuroblast proliferation  
cardiac muscle tissue growth  
negative regulation of smoothed signaling pathway  
heart growth  
cardiac epithelial to mesenchymal transition  
detection of mechanical stimulus involved in sensory perception  
ventricular cardiac muscle tissue morphogenesis  
ventricular cardiac muscle tissue development  
positive regulation of phospholipid metabolic process  
oligodendrocyte differentiation

## eGenes associated with the 20 most significant fQTLs [8 cell analysis]

CLDN11

Multiple Sclerosis

DPYD

Colorectal Neoplasms

Carcinoma, Hepatocellular

Neutropenia

**Peripheral Nervous System Diseases**

**Autistic Disorder**

Head and Neck Neoplasms

Lung Neoplasms

Pancreatic Neoplasms

**Schizophrenia**

Colonic Neoplasms

Leukoencephalopathies

Neoplasm Metastasis

Obesity

**Autism Spectrum Disorder**

Breast Neoplasms

Nervous System Diseases

Stomach Neoplasms

Dihydropyrimidine Dehydrogenase Deficiency

**Language Development Disorders**

Purine-Pyrimidine Metabolism, Inborn Errors

eGenes associated with the 20 most significant fQTLs [8 cell analysis]

GPR153

**Schizophrenia**

IL1RAP

Liver Diseases

MSX1

Craniofacial Abnormalities  
Tooth Abnormalities  
Wolf-Hirschhorn Syndrome  
Anodontia  
Witkop syndrome  
Cleft Palate  
Stomach Neoplasms  
Orofacial Cleft 5

eGenes associated with the 20 most significant fQTLs [8 cell analysis]

PLD1

Infarction, Middle Cerebral Artery  
Cardiomegaly

RGS12

**Schizophrenia**

SST

Hypokalemia  
**Ischemia**  
**Mood Disorders**  
Vipoma  
Multiple Organ Failure  
**Seizures**  
Barrett Esophagus  
Neoplasms  
Abortion, Spontaneous  
Esophageal Neoplasms  
Pancreatic Neoplasms  
Pancreatitis  
Endometriosis  
Esophageal and Gastric Varices

## eGenes associated with the 20 most significant fQTLs [8 cell analysis]

TF

Adenocarcinoma of lung  
Hepatitis  
Paratuberculosis  
Anemia, hypochromic microcytic  
Iron Overload  
Mammary Neoplasms, Animal  
**Alcoholism**  
Carcinoma  
Congenital atransferrinemia  
Fatty Liver  
**Alzheimer Disease**  
Drug-Induced Liver Injury  
Echinococcosis  
Nephrotic Syndrome  
Inflammation  
Neoplasm Invasiveness  
**Restless Legs Syndrome**  
**Autistic Disorder**  
Mammary Neoplasms, Experimental  
Mesothelioma, Malignant  
Obesity



## Summary data on fQTLs calculated from 24 cell types

Cell_type	num_fQTLs	min_bonf_corrected_p_val	max_bonf_corrected_p_val
Ex6	15664	5.84E-23	0.049929385
Fetal-replicating	12412	2.29E-18	0.049950551
In5	9444	2.16E-29	0.049976051
In1	4701	1.02E-58	0.049933975
In4	4133	3.10E-25	0.049991607
Fetal-quiescent	3958	5.55E-13	0.049987782
In3	2482	1.01E-17	0.049816673
Ex7	2257	6.80E-21	0.049925943
Ex2	2074	2.55E-52	0.049865889
Ex8	1369	1.78E-46	0.049889222
OPC	1252	1.31E-53	0.048778289
In8	902	1.73E-19	0.049814123
Ex4	380	4.37E-22	0.04993066
In6	113	1.76E-14	0.048444104
Ex3	58	5.79E-08	0.04883592
Astrocytes	42	4.33E-06	0.042957397
Microglia	23	1.22E-05	0.048852113
Ex5	9	3.96E-09	0.047241623
Oligodendrocytes	6	4.15E-07	0.038137404
Ex1	0	--	--
In2	0	--	--
In7	0	--	--
Endothelial	0	--	--
Neuron	0	--	--

# Stats on fQTLs w/SNVs that coincide with trans-eQTLs [24 cells analysis]

439/457 genes have all needed data (ENSG ID, TSS, etc)

24 cell types				
p_val_thresh	tot # matches	# unq matched SNVs	# unq fQTL SNVs	fract unq fQTL SNVs that match eSNVs
0.05	520995	<b>22885</b>	41465	<b>0.552</b>
0.1	637069	26639	47223	0.564
0.2	777733	30850	53569	0.576
0.3	873281	33529	57341	0.585

\* P-values are Bonferroni-corrected (for both fQTLs and trans-eQTLs)

## Trans-eGenes associated with the 20 most significant fQTLs (linked through common SNV) [24 cell analysis]

Had to take the top 53 most significant fQTLs to build a list of 20 unique genes – ie, there's considerable over-representation of certain genes among the most significant matches

ATP10A	ATPase phospholipid transporting 10A (putative)
BRIP1	BRCA1 interacting protein C-terminal helicase 1
CAMK2A	calcium/calmodulin dependent protein kinase II alpha
CENPF	centromere protein F
DPYSL3	dihydropyrimidinase like 3
ENPP2	ectonucleotide pyrophosphatase/phosphodiesterase 2
EYA1	EYA transcriptional coactivator and phosphatase 1
HAS2	hyaluronan synthase 2
MDGA1	MAM domain containing glycosylphosphatidylinositol anchor 1
MEX3A	mex-3 RNA binding family member A
MSX1	msh homeobox 1
MYB	MYB proto-oncogene, transcription factor
NMBR	neuromedin B receptor
PBK	PDZ binding kinase
PRDM8	PR/SET domain 8
RELN	reelin
SLC10A4	solute carrier family 10 member 4
STK32B	serine/threonine kinase 32B
TESC	tescalcin
VAV3	vav guanine nucleotide exchange factor 3

## A general note on the number of ways to compute QTLs (at least $2^5 = 32$ )

normalized vs. **non-normalized** [2]

w/peer or **w/o** peer [2]

cis or **trans** [2]

calculate significance via **Bonferroni** on nominal run or FDR w/permutations [2]

significance thresholds (**.05**, .10) [2]

