fQTLs btwn 9 phenotypes & 1452 individuals (5,312,628 variants)

Overall Scheme

Step 1: Normalize the cell fraction matrix (Quantile normalization, Inverse quantile normalization, etc)
+ results from non-normalized matrix also available for comparisons

Step 2: Run a nominal pass (ie, calculate QTLs in the usual way and report nominal p-values)

```
Ex6 chr1 1 1:5016315 chr1 5016315 9.80619e-06 -1 0.115741
Ex8 chr1 1 1:5016315 chr1 5016315 9.35944e-06 -1 0.116003
Ex6 chr1 1 1:5018078 chr1 5018078 7.61258e-06 -1 0.117155
Ex4 chr1 1 1:5030204 chr1 5030204 1.8558e-07 -1 -0.13629
Ex6 chr1 1 1:5030204 chr1 5030204 9.26592e-09 -1 0.150007
Ex8 chr1 1 1:5030204 chr1 5030204 1.8623e-06 -1 0.124741
In1 chr1 1 1:5030204 chr1 5030204 7.69179e-07 -1 0.129287
```

1. Phenotype ID
2. Phenotype chrID
3. Phenotype start
4. Variant ID
5. Variant chrID
6. Variant position
7. Nominal P-value of association
8. Dummy variable.
9. Regression slope

Step 3: Controlling for multiple tests: Bonferroni-corrected p-values (Tot # tests: 9 * 5,312,628 ~ 48 million)

#Cell_Type	Chr_of_SNV	Locus_of_SNV	Nominal_p_val	Bonferroni_corrected_p_val	Regression_slope
Ex5	chr1	7706417	4.50415e-14	2.15359860656e-06	0.196259
Ex5	chr1	7706563	1.25981e-14	6.02361169261e-07	0.200447
Endothelial	chr1	7706563	2.73318e-12	0.000130683317373	0.182082
Ex5	chr1	10822171	3.00088e-10	0.0143483032014	-0.164316
Astrocytes	chr1	10822171	3.77327e-10	0.0180413818682	0.163399
Ex5	chr1	24032615	8.81277e-10	0.0421370717936	-0.15996
Astrocytes	chr1	24336314	2.20708e-15	1.05528555056e-07	0.206027
Astrocytes	chr1	24336437	2.30179e-15	1.10056986037e-07	0.205894
Ex5	chr1	27977923	6.40507e-16	3.06249788016e-08	0.209895
Ex5	chr1	27980793	7.96136e-15	3.80661696487e-07	0.201933

9 cell types included

- Ex1
- Ex3
- Ex4
- Ex5
- In6
- Astrocytes
- Endothelial
- Microglia
- Oligodendrocytes

# of significant* fQTLs produced from the pre-normalized cell fractions matrix: 508

# of significant* fQTLs produced from the normalized cell fractions matrix: 751

** Significant if Bonferroni-corrected p-value < 0.05*

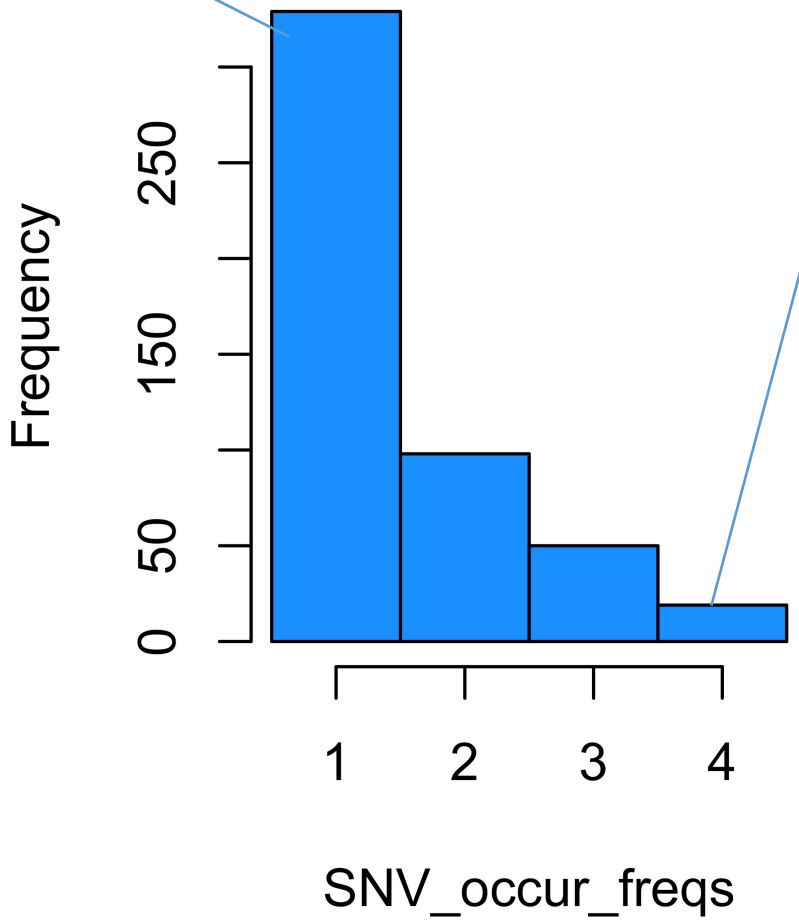
Summary Stats Derived from the **Pre-Normalized** Matrix			
Cell_type	num_fQTls	min_bonf_corrected_p_val	max_bonf_corrected_p_val
Ex4	318	1.38E-08	0.049120399
Astrocytes	15	2.20E-12	0.046619076
Microglia	33	3.78E-05	0.045288804
Endothelial	45	1.25E-08	0.048274576
Ex3	66	2.07E-07	0.045465189
Ex5	27	7.16E-06	0.030902537
Ex1	4	0.001027295	0.026625654
In6	0	-	-
Oligodendrocytes	0	-	-

Derived from **Normalized** Matrix			
Cell_type	num_fQTls	min_bonf_corrected_p_val	max_bonf_corrected_p_val
Ex5	476	5.64E-24	0.048395544
Endothelial	144	3.09E-11	0.049015209
Astrocytes	91	4.03E-11	0.048337211
Ex4	37	1.13E-05	0.043376067
In6	2	0.001328392	0.003109059
Ex3	1	0.048427101	0.048427101
Ex1	0	-	-
Microglia	0	-	-
Oligodendrocytes	0	-	-

To what extent do certain genomic variants re-occur among the set of fQTLs?

(# of available SNVs: ~5.3 million)

Among 751 fQTLs, there were 329 distinct SNVs such that each SNV was an "fSNV" for 1 cell type -- ie, these SNVs were 'weaker cell fraction determinants'



Among these 751 fQTLs, there were 19 distinct SNVs such that each SNV was an "fSNV" for 4 different cell types -- ie, these SNVs were 'very powerful cell fraction determinants'

8/19 in Chr 1 (all around same locus)
all around locus 194182229
8/19 in Chr 19:
6 around locus 47888815
2 around locus 8277599

**showing results derived from normalized dataset*

Do recurrent ”fSNPs” exert stronger effects (as measured by regression slopes)?

8/19 in Chr 1 (all around same locus)
all around locus 194182229
8/19 in Chr 19:
6 around locus 47888815
2 around locus 8277599