## 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	1. Phenotype ID
Ex8	chr1	1	1:5016315	chr1	5016315	9.35944e-06 -1 0.116003	2. Phenotype chrlD 3. Phenotype start
Ex6	chr1	1	1:5018078	chr1	5018078	7.61258e-06 -1 0.117155	4. Variant ID
Ex4	chr1	1	1:5030204	chr1	5030204	1.8558e-07 -1 -0.13629	5. Variant chrID
Ex6	chr1	1	1:5030204	chr1	5030204	9.26592e-09 -1 0.150007	6. Variant position
Ex8	chr1	1	1:5030204	chr1	5030204	1.8623e-06 -1 0.124741	7. Nominal P-value of association
In1	chr1	1	1:5030204	chr1	5030204	7.69179e-07 -1 0.129287	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	# of significant* fOTLs produced		
Ex1	from the pre-normalized cell		
Ex3	fractions matrix: 508		
Ex4			
Ex5			
In6	# of significant* fQTLs produced		
Astrocytes	from the normalized cell fractions		
Endothelial	matrix: 751		
Microglia			
Oligodendrocytes			

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

Summary Stats Derived from the Pre-Normalized Matrix					
<u>Cell_type</u>	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						
<u>Cell_type</u>	<u>num_fQTIs</u>	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)



SNV\_occur\_freqs

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



Frequency

regression slopes (for Chr 1 hotspot)