

To compute fQTLs:

- + cell fractions file used as is
- + sheared up genome into 250 pieces (rapid calc)
- + computed fQTLs in a 'cis' manner in order to include covariates (next slide)
- + ran a 'hacked' version of the cis search (w/covariates) to identify fQTLs
 - sanity checks in QTLtools – effects of varying window sizes
 - cell types added as "genes" to each chromosome
 - set cis-window length to exceed the length of the chromosome
 - merge results for each cell type across chromosomes
- + Bonferroni correction after nominal pass

Filter input files to include only adult samples w/genotype data and genotype PCs: 1362 samples

1) Genotype file

2) Covariates file

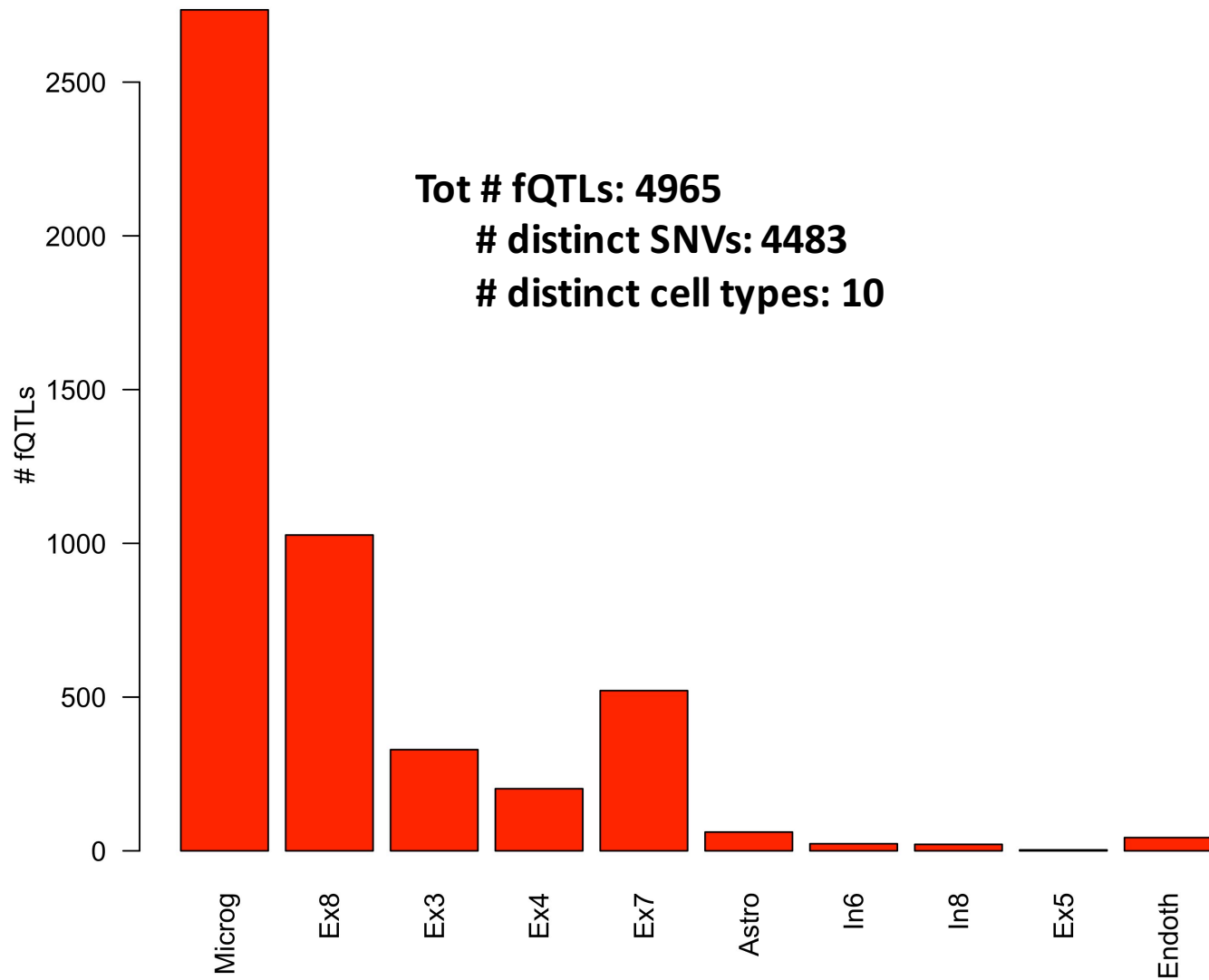
- 3 genotype PCs
- gender
- disease status
- study (GTEx, Brainspan, Bipseq, etc)
- NOT included as covariates:
 - + age
 - + PEER factors (not appropriate for cell fractions)
 - + genotype PCs for 25 GTEx samples

3) Cell fractions (ie, the phenotype file)

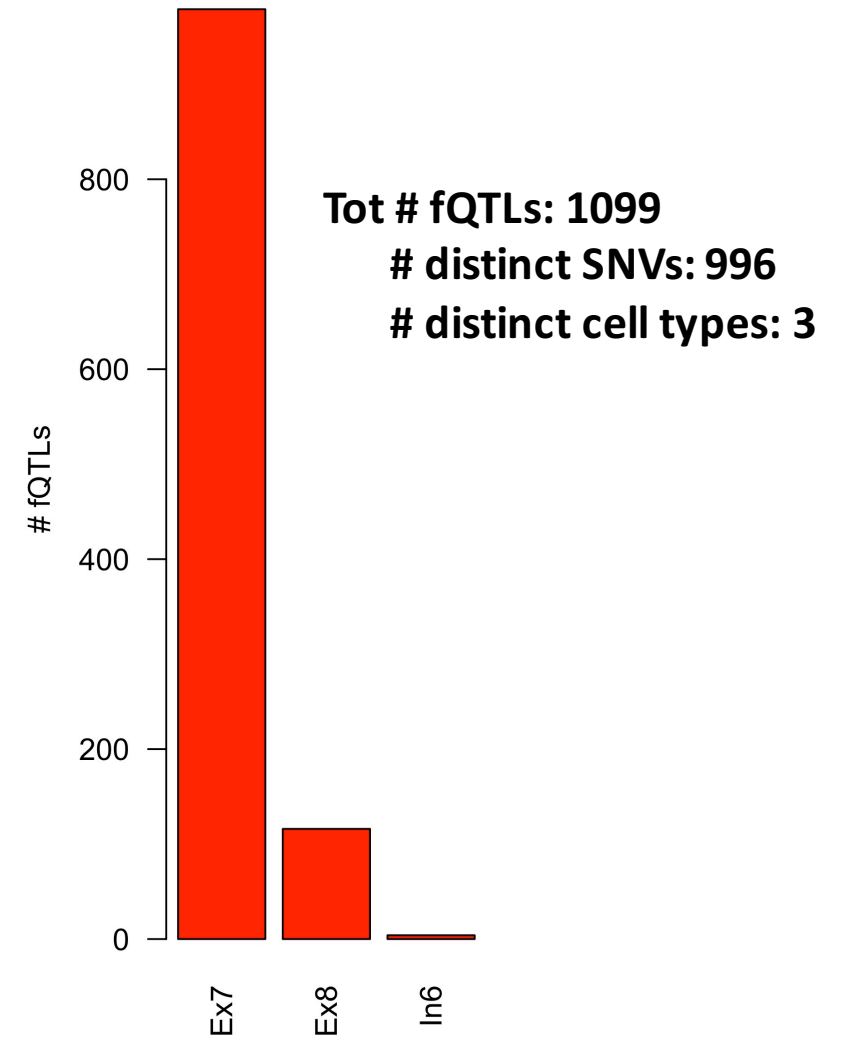
includes only the 10 selected cell types on which we've previously decided

- Ex3
- Ex4
- Ex5
- Ex7
- Ex8
- In6
- In8
- Astrocytes
- Endothelial
- Microglia

fQTLs without covariates



fQTLs w/covariates



Pending (in order)

- + if necessary: improvements to current fQTLs identification pipeline (w/covariates)
- + re-calc (based on updated fQTLs): residual-QTLs on 440 biomarker genes (first trans, then cis)
- + SNV overlaps accr. QTL types
- + adaptive thresholding
- + could we do the fQTL like height?