



Figure S4 Effects of *cis*-genetic variants (those within 10kb of the TSS) on allelic imbalance. Variants binned by minor allele frequency in GTEx. Estimates from combined-tissue ASE (reads spanning each ASE site in a single individual are summed, regardless of the tissue in which they were sampled) are shown as large points. Small points represent estimates from 100 permutations of ASE measurements across individuals within a gene. The estimated singleton effect size from real data is significantly greater than those from the permutations. However, correlation in the allele frequency spectra at a locus across individuals clearly contributes to our effect size estimates. One interpretation of this is that genes vary in their level of constraint on expression. This could lead to subsets of genes with different frequency spectra and different ASE profiles.