

Figure S1 Comparison of eQTLs called using forward-stepwise mapping in an ascertainment set of 800 individuals, effect sizes estimated in 122 individuals. Data are whole blood RNA sequencing from the DGN cohort. *cis*-eQTLs were called in a 100kb window centered on the TSS of autosomal, protein-coding genes. Effect sizes are polarized relative to the ancestral allele. (A) eQTL effect sizes (estimated in the ascertainment set) as a function of eQTL minor allele frequency. eQTLs that we were uniformly powered to detect across allele frequencies (estimated effect greater than the minimum effect size estimated for eQTLs with MAF < 0.02) shown in black, others (including especially rare eQTLs, MAF < 0.02) in grey. Loess-fits of minor allele frequency vs eQTL effect size (for well-powered expressionincreasing and -decreasing eQTLs, respectively) are shown in blue. (B) eQTL effect sizes (estimated in the validation set) as a function of eQTL minor allele frequency. Only eQTLs that we were well-powered to detect across allele frequencies and with MAF > 0.02 are plotted. Points are colored by the direction of the eQTL effect in the ascertainment set; expression-increasing eQTLs (derived allele predicted to increase expression) shown in dark blue, expressiondecreasing eQTLs in light blue. Loess-fits of minor allele frequency vs eQTL effect size (for expression-increasing and -decreasing eQTLs, respectively) are shown in black. (C) Correlation between effect sizes estimated in the larger, ascertainment sample (x-axis) and the smaller, validation sample (y-axis). eQTLs that we were uniformly powered to detect across allele frequencies (estimated effect greater than the minimum effect size estimated for eQTLs with MAF < 0.02) shown in black, others (including rare eQTLs, MAF < 0.02) in grey. y = x line is shown in black, least-squares regres sion line shown in blue.