SUPPLEMENTAL FIGURES

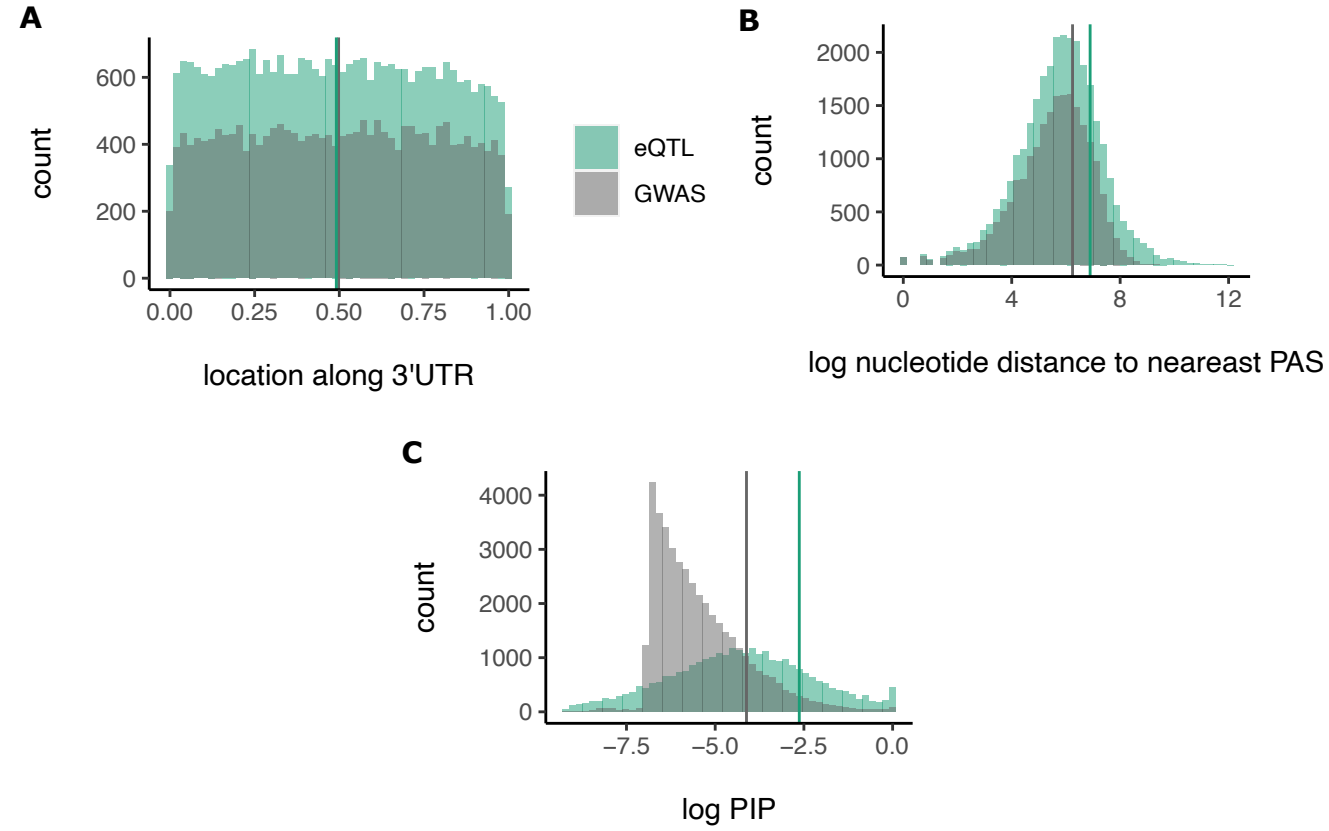

Figure S1: Comparison of eQTL variant and GWAS hit distribution along 3'UTR (A), proximity to nearest polyA signal (B), and PIPs (C).

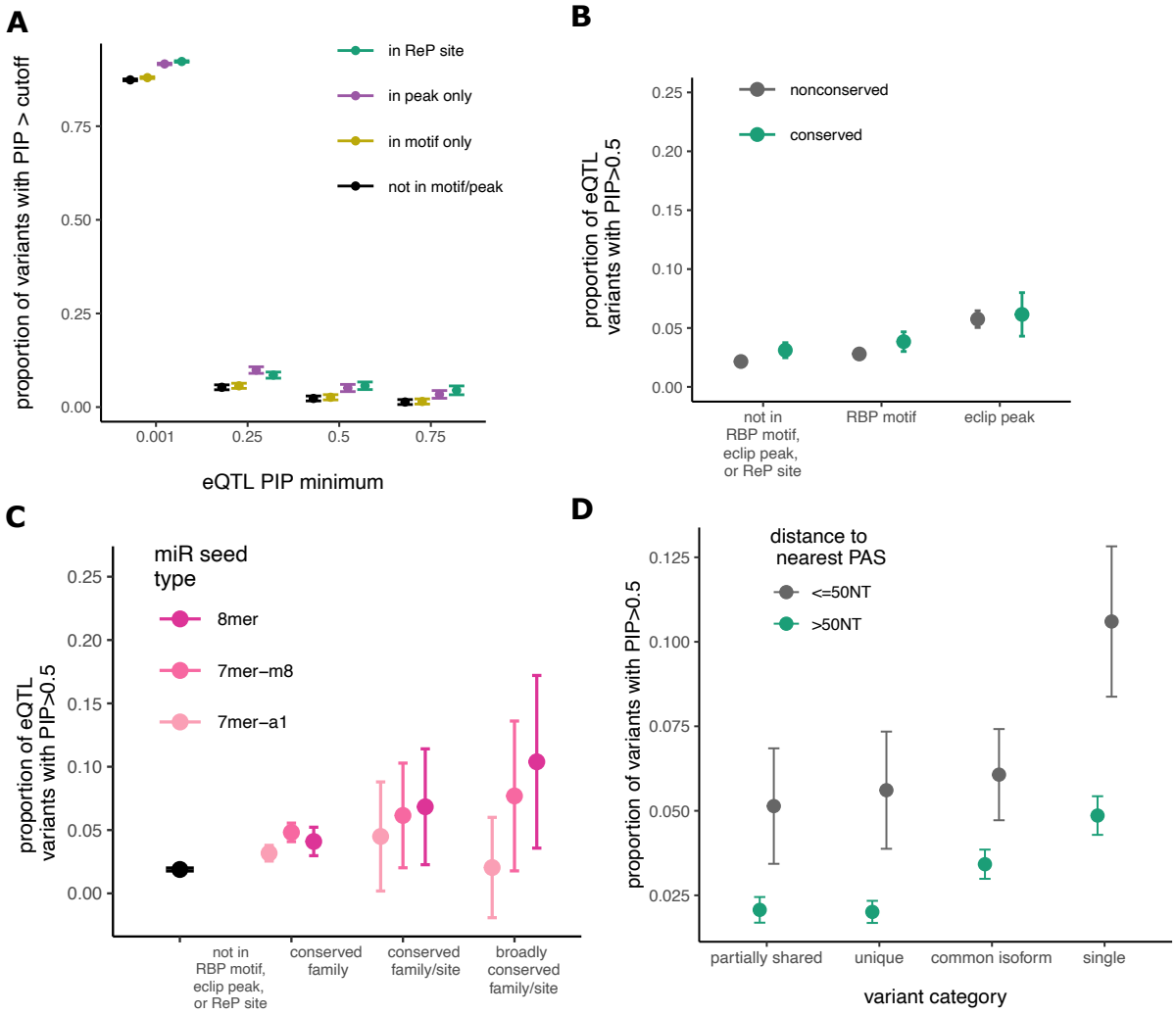

Figure S2: eQTL findings are robust even with a more stringent summary statistic PIP threshold. A. Proportion of eQTLs with PIP greater than a minimum cutoff for variants not in RBP motifs or eCLIP peaks compared to variants in RBP motifs, eCLIP peaks, and ReP sites, with 95% confidence intervals. **B.** Fraction causal (proportion of eQTLs with PIP>0.5) for variants not in RBP motifs or eCLIP peaks compared to variants in RBP motifs or eCLIP peaks. **C.** Fraction causal for variants not in miRNA sites compared to variants in miRNA sites with increasing predicted seed strength. **D.** Fraction causal for eQTL variants in genes with various numbers of canonical alternatively polyadenylated (APA) isoforms.

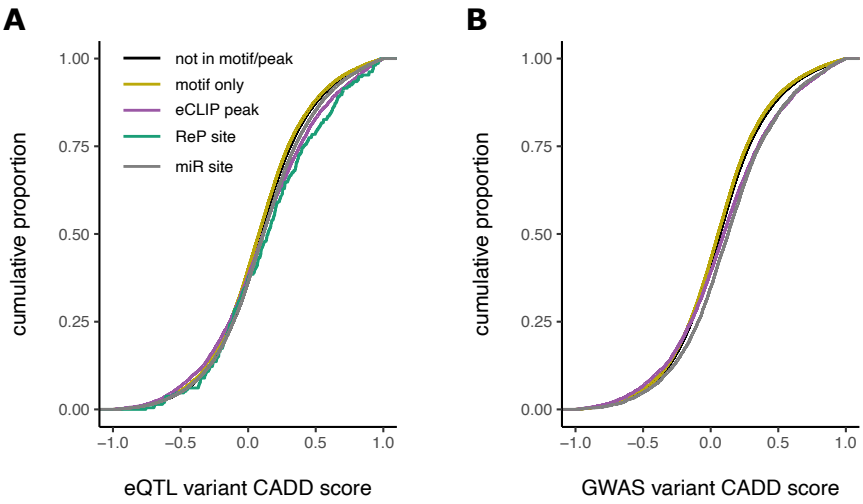

Figure S3: Variants in putative regulatory elements have higher CADD scores. Comparison of raw combined annotation dependent depletion (CADD) score distributions for eQTLs (A) or GWAS hits (B) in various putative regulatory elements versus controls.

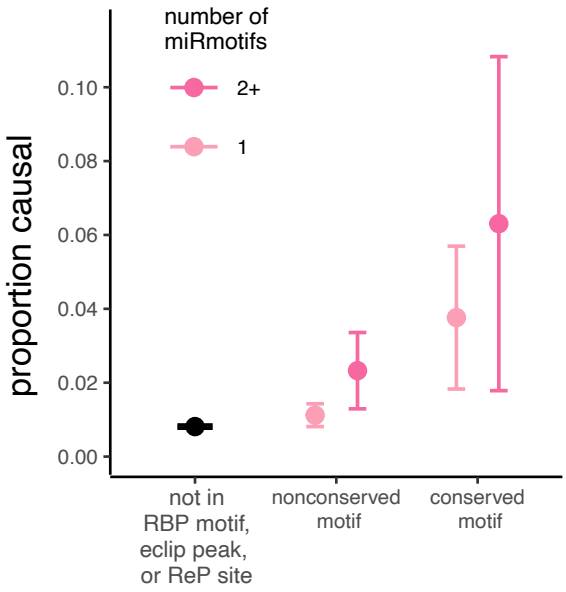

Figure S4: Trend towards higher PIP for variants predicted to disrupt more than one miRNA site. Fraction causal for GWAS variants not in miRNA sites compared to variants in increasing number of sites.

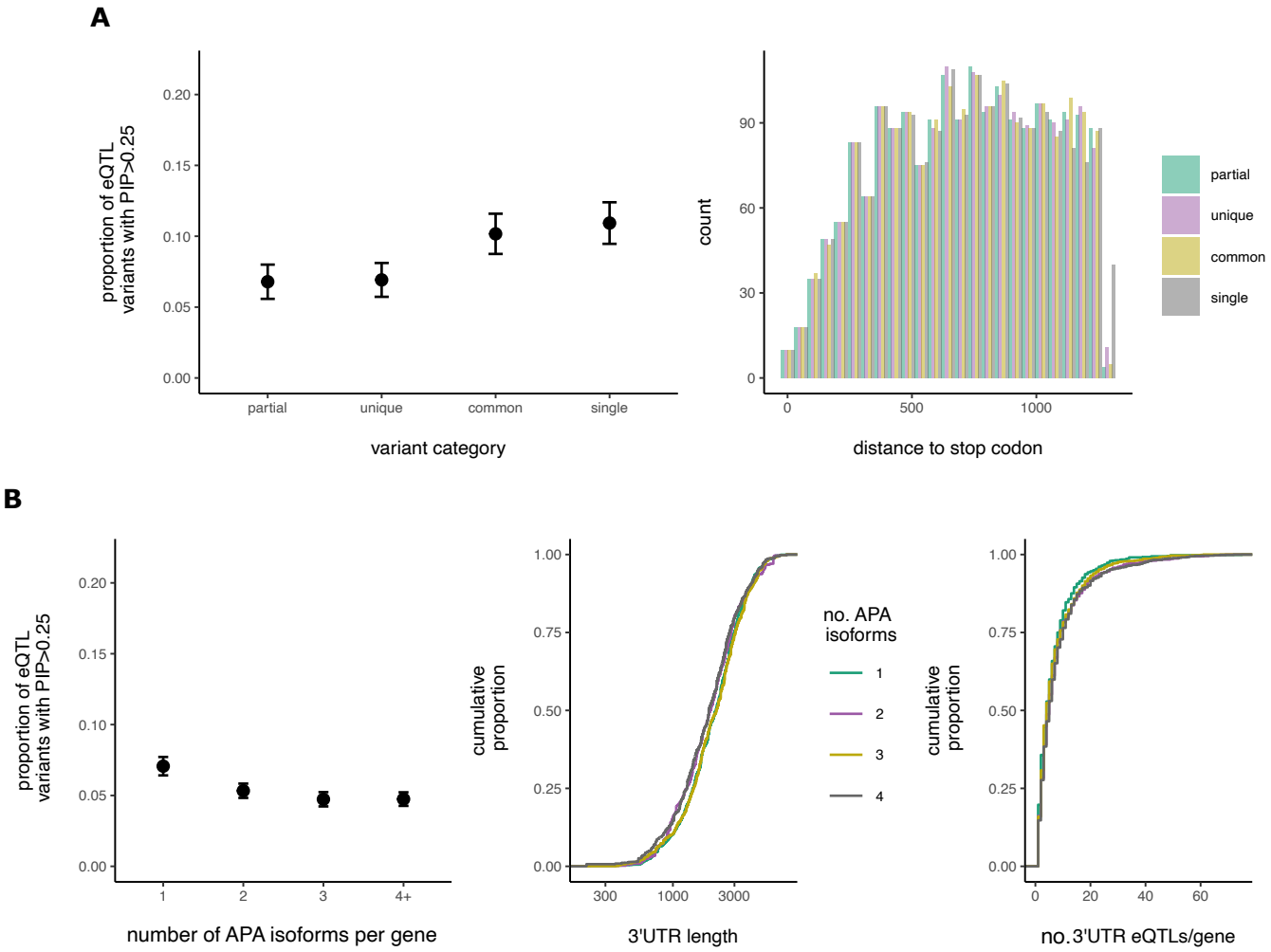

Figure S5: eQTL findings are not due to stop proximity, 3'UTR length, or number of eQTLs per gene. A. Fraction causal (proportion of eQTL variants with PIP greater than 0.25) for variants in various 3'UTR regions (left), after matching distance to canonical stop codon (right). **B.** Fraction causal for eQTL variants in genes with various numbers of canonical alternatively polyadenylated (APA) isoforms (left) after matching gene 3'UTR length (middle). On right is the distribution of number of eQTLs per gene for genes with varying isoform numbers.

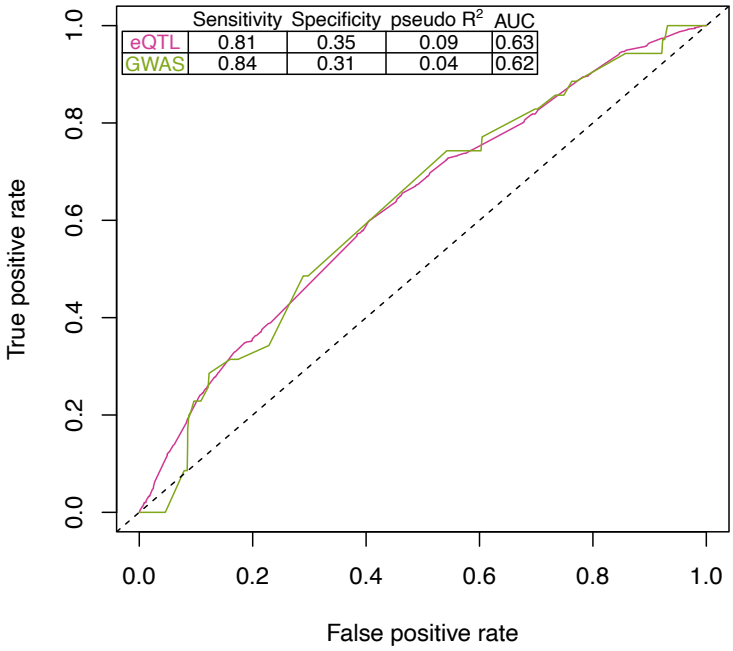

Figure S6: Performance of generalized linear models. Logistic regression analysis was performed to predict GWAS and eQTL variants (PIP>0.5). A variant was predicted to be an eQTL or GWAS hit if its log-odds was greater than 0.01 (eQTL) or 0.0075 (GWAS). These thresholds maximized sensitivity and specificity. Goodness of fit was assessed via Hosmer-Lemeshow Test with a chi squared of 1.0204 and p-value of 0.9981 for the eQTL model and a chi squared of 13.262 and p-value of 0.1032 for the GWAS model.

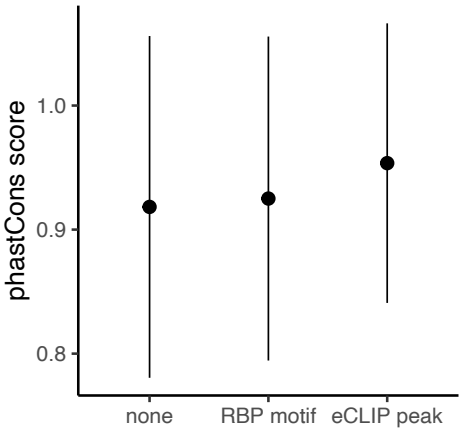

Figure S7: enrichment for pathogenic variants in regulatory elements is not solely due to conservation. Shown is mean phastCons score with standard deviation for variants in each category.