

Description of Additional Supplementary Files

Supplementary Data 1. The number of puQTLs and promoters across 49 tissues. Supplementary Data 1 provides information on the number of puQTLs and promoters across 49 tissues.

Supplementary Data 2. The promoters with multiple puSNPs across 49 tissues. Supplementary Data 2 presents data on the promoters that have multiple puSNPs across the 49 tissues. N presents the number of puSNPs per promoter.

Supplementary Data 3. The metadata of HiChIP data matching with tissues. The metadata of HiChIP data is recorded in Supplementary Data 3, detailing the specific tissues and cell types to which the data corresponds, as well as the corresponding GEO accession number.

Supplementary Data 4. The chromatin interactions and distance between puQTLs and promoters. The chromatin interactions and distance between puQTLs and promoters are described in Supplementary Data 4. This table also includes the number of puQTLgene pairs verified by HiChIP loops and categorizes them based on distance and verification status.

Supplementary Data 5. Genes associated with paQTLs classified into five groups. Supplementary Data 5 classifies genes associated with paQTLs into five groups and provides information on their pairing with promoters, effect directions, and effect size.

Supplementary Data 6. The overlapping between puSNP-gene pairs and paSNPgene pairs. Supplementary Data 6 presents the overlapping puSNP-genes and paSNPgenes, categorized into four groups.

Supplementary Data 7. The different pairing and regulation patterns of puSNP-gene and paSNP-gene pairs. Supplementary Data 7 describes the different pairing and regulation patterns observed in puQTL and paQTL pairs, noting the number of overlaps with the same or different promoters and effect directions.

Supplementary Data 8. The overlap between puQTL-genes and eQTL-genes. The overlap between puQTL-genes and eQTL-genes is presented in Supplementary Data 8.

Supplementary Data 9. TF binding motifs disrupted by puQTLs. Supplementary Data 9 lists the TF binding motifs disrupted by puQTLs in each tissue, along with the odds ratio and significance of the disruption.

Supplementary Data 10. List of GWAS complex traits examined in this study. Supplementary Data 10 provides a comprehensive list of the GWAS complex traits examined in this study, including sample size, population, category, and PubMed ID.

Supplementary Data 11. List of puQTLs colocalized with human trait GWAS signals. Supplementary Data 11 presents the colocalization of puQTLs with complex traits identified through GWAS. The table includes essential information such as the promoter ID, gene name, trait, tissue, and five posterior probabilities. PP0 represents the null model in which neither the

GWAS trait nor the promoter usage exhibit a genetic association in the region. PP1 signifies that only the GWAS trait exhibits a genetic association in the region. PP2 indicates that only the promoter usage displays a genetic association. PP3 denotes that both the GWAS trait and the promoter usage are associated, but with two independent SNPs. PP4 indicates that both the GWAS trait and the promoter usage are associated with a shared single causal variant.