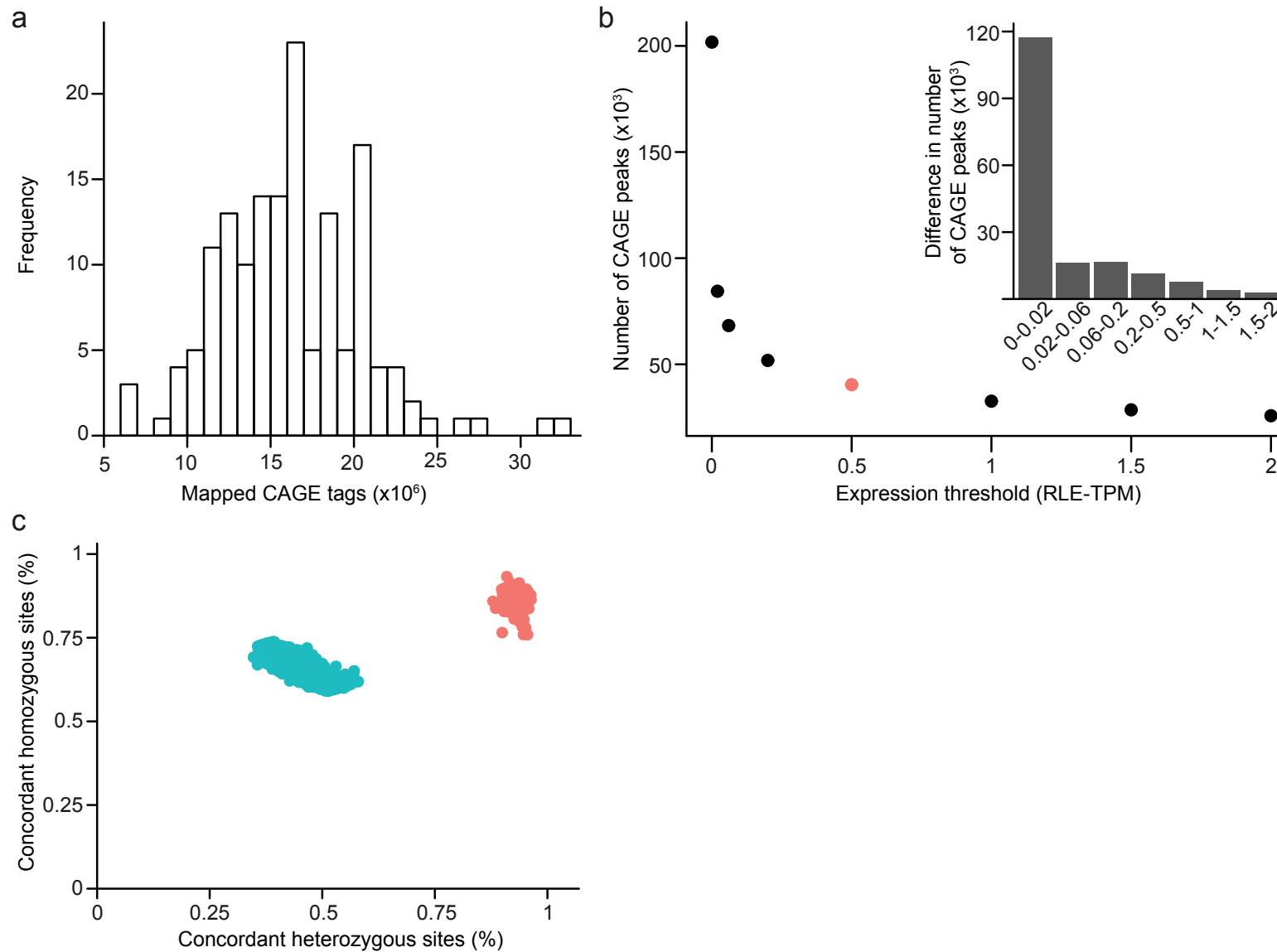
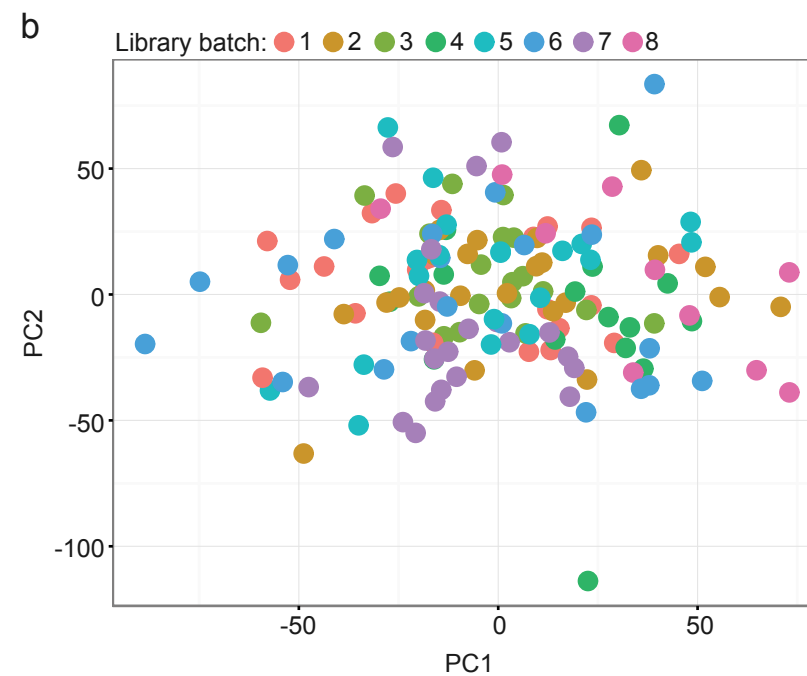
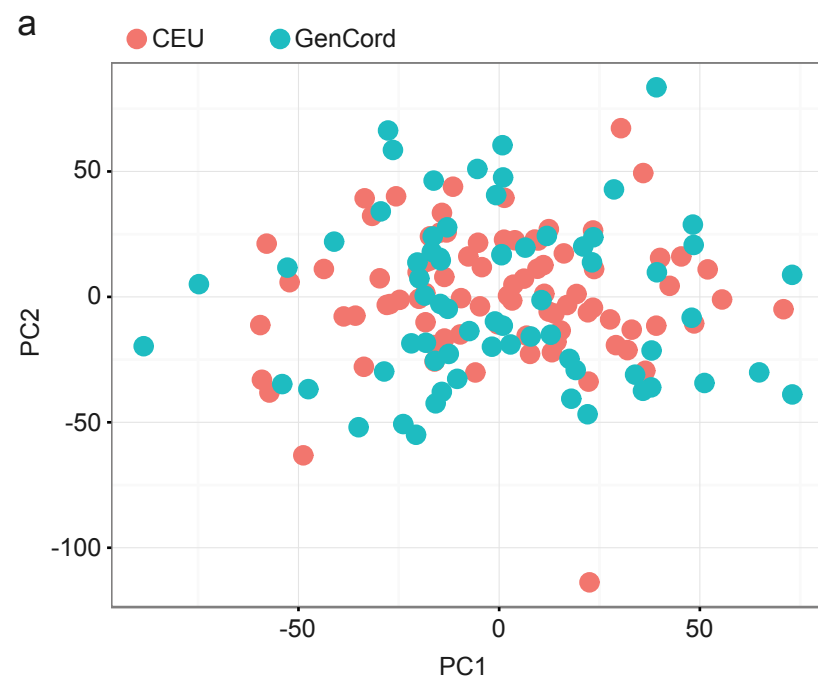
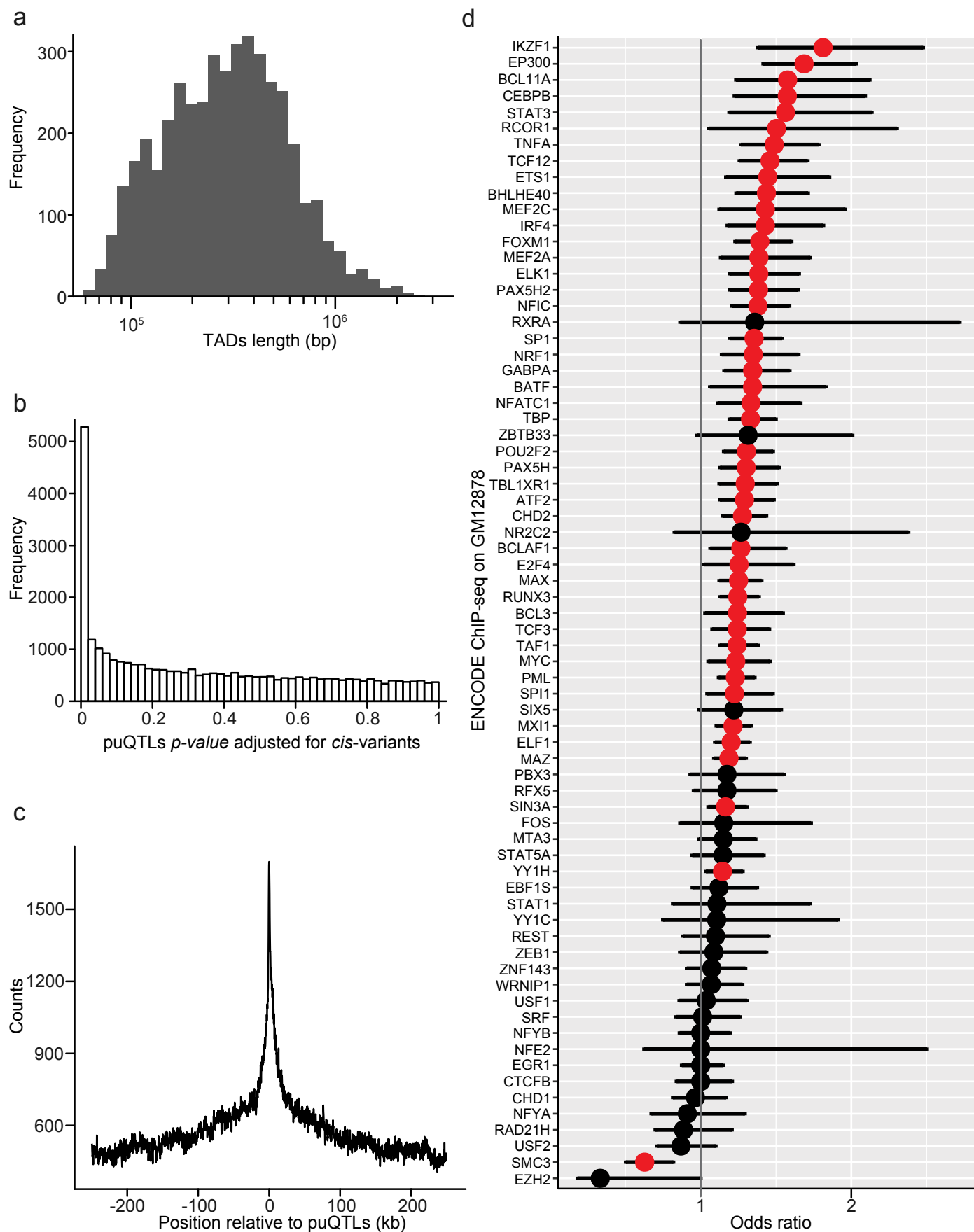




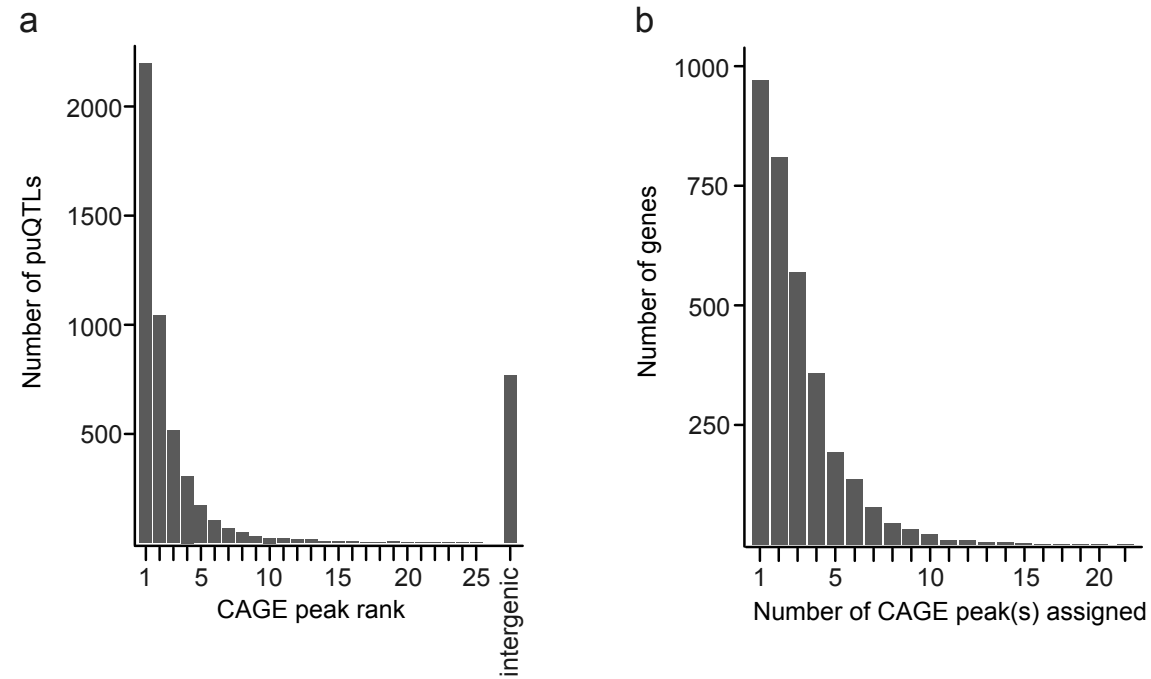
**Supplementary Figure 1: Sequencing statistics and quality control.** (a) Sequencing depth is shown as a histogram of single mapped CAGE tags (MapQ>20) for 154 individuals. (b) Distribution of CAGE peaks (including X and Y chromosomes) passing different expression thresholds. The 0.5 RLE-TPM threshold used in the study is highlighted in red. The bar-plot shows the number of CAGE peaks lost between consecutive thresholds. (c) *MBV* output (Methods) allelic consistency for heterozygous (x-axis) and homozygous (y-axis) variants, between genotype and CAGE tag sequences are plotted. Red dots indicate proper identity match of sequencing with expected genotypes. As control, we show consistency levels for non-matching samples (blue dots).



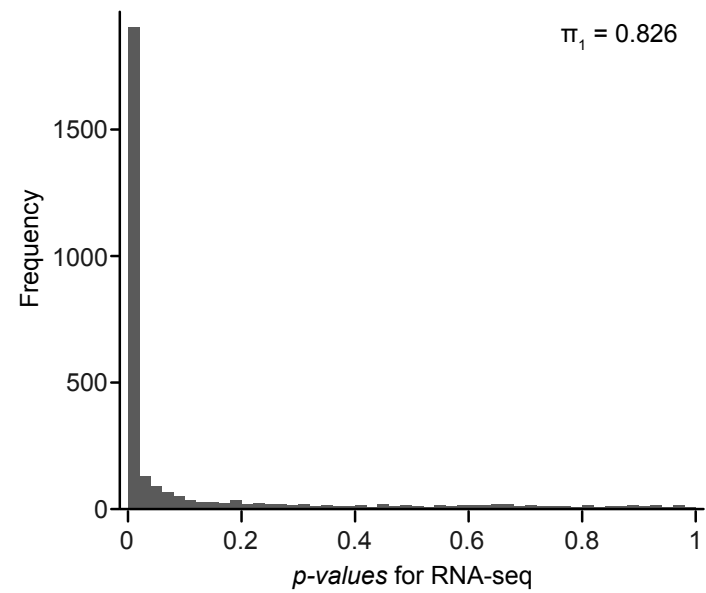
**Supplementary Figure 2: Data stratification.** Projection onto the first two principal components of the normalized data, residualized for the first 3 PCs derived from genotype data and the first 20 PCs of the promoter expression values, labelled for **(a)** the two cohorts used in the study and **(b)** for batches of library preparations.



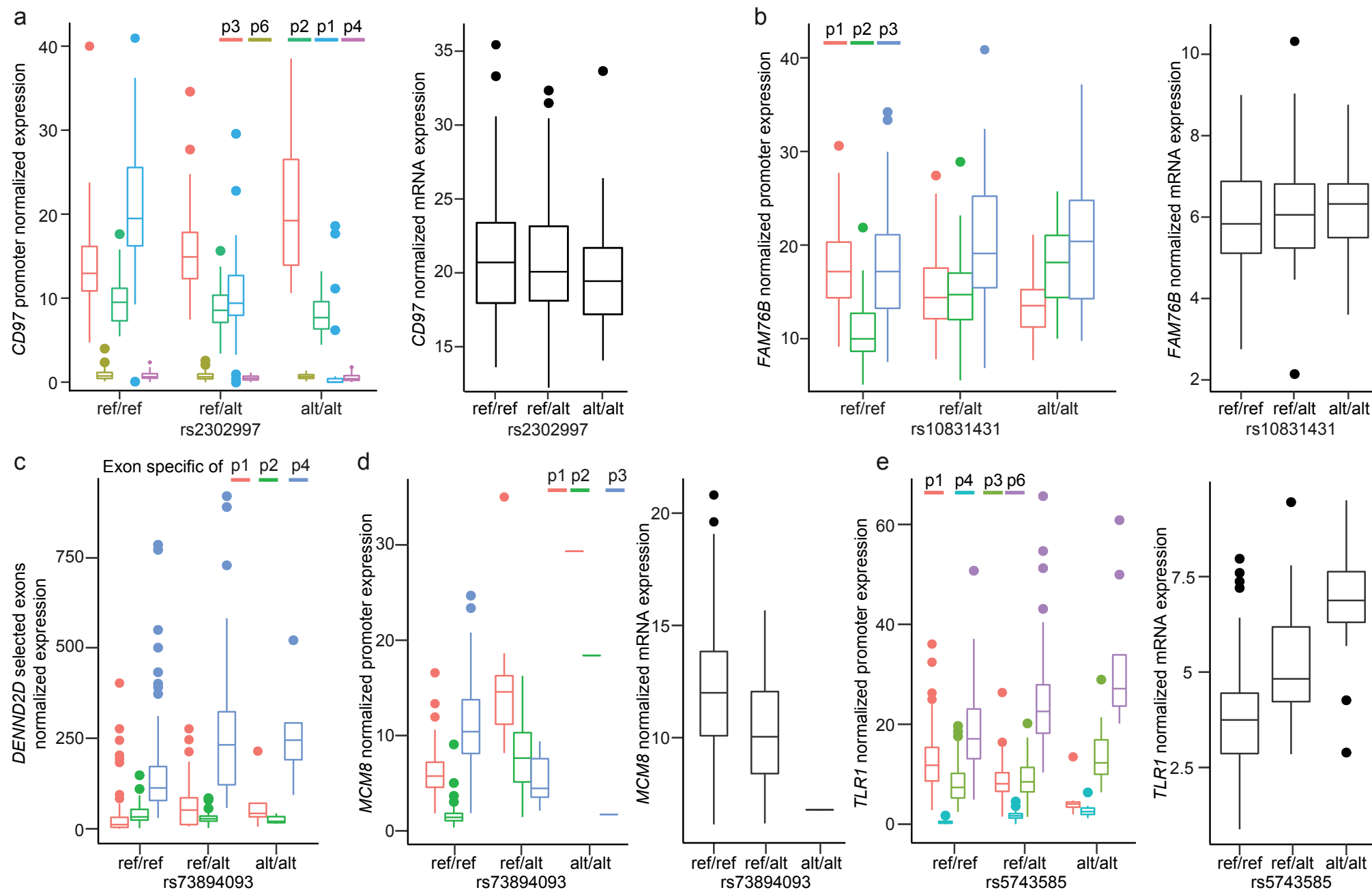
**Supplementary Figure 3: puQTLs mapping and enrichment analyses.** (a) Distribution of TADs length (b) Distribution of *p*-values adjusted for number of tested *cis*-variants associations with promoters. (c) Total counts of DNase I hypersensitivity signal (GM12878 cells, ENCODE data) for the 500 kb region flanking puQTLs. (d) Enrichment of transcription factor ChIP-seq signal (GM12878 cells, ENCODE data) at puQTL sites. Red dots indicate values passing the significance threshold of 5% FDR and bars show 95% confidence intervals.



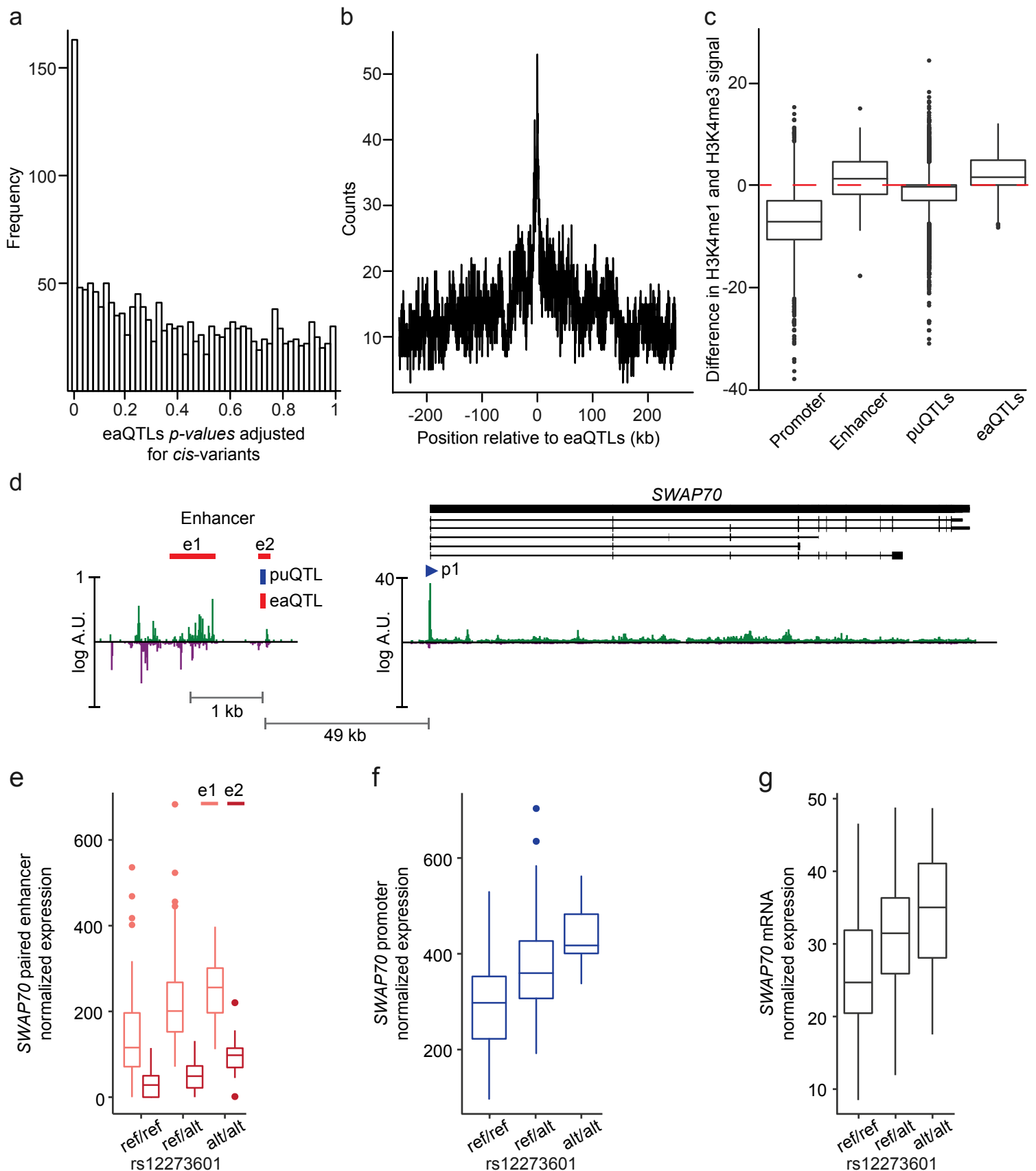
**Supplementary Figure 4: FANTOM CAGE peaks annotation.** (a) Distribution of puQTLs for the CAGE peak rank and (b) number of CAGE peaks per gene.



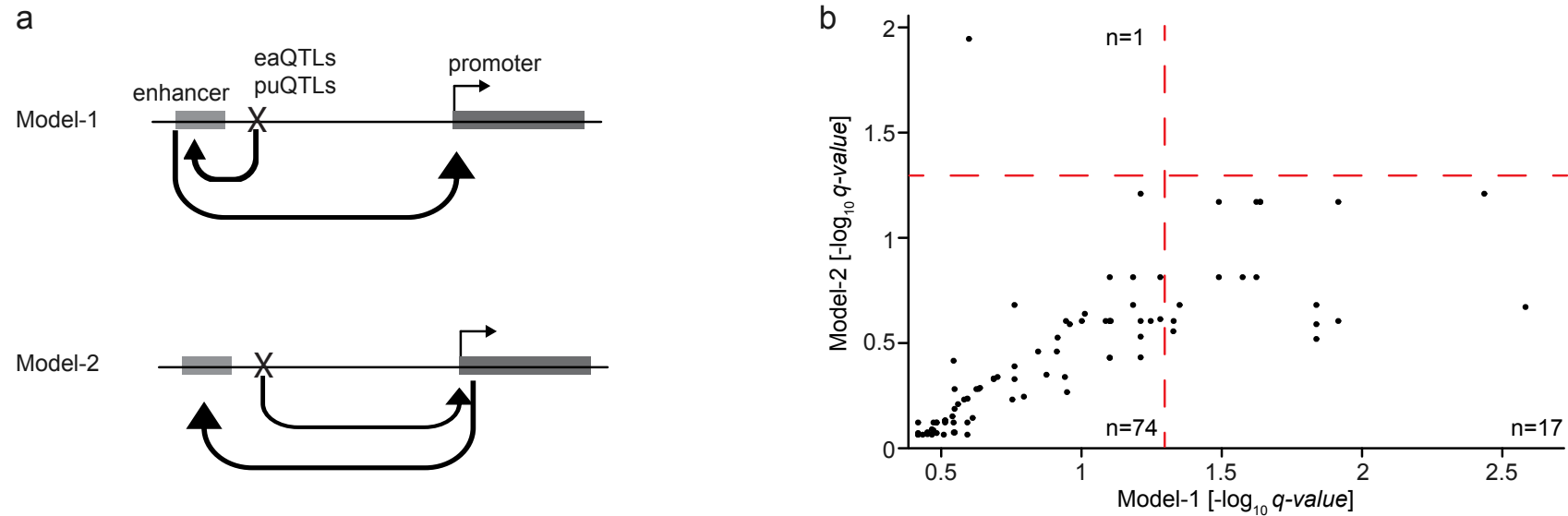
**Supplementary Figure 5:  $\pi_1$  statistic** for puQTLs replicating eQTLs calling for mRNA levels measured from RNA-seq data.



**Supplementary Figure 6:** Normalized expression for promoters (colored box plots) and mRNA levels (black box plots) relative to puQTL genotypes are plotted for *CD97* (a) and *FAM76B* (b) both belonging to the puQTLs associated gene group-3 without effect on mRNA levels. (c) Quantification of exons specific to different *DENND2D* isoforms are plotted for the different puQTL genotypes. Normalized expression for promoters and mRNA levels relative to puQTLs genotypes are plotted for *MCM8* (d) and *TLR1* (e) both belonging to the puQTLs associated gene group-3, with observed eQTL effect.



**Supplementary Figure 7: eaQTLs mapping and integration with puQTLs.** (a) Distribution of  $p$ -values adjusted for number of tested  $cis$ -variants associated with enhancers. (b) Total counts of DNase I hypersensitivity signals (GM12878 cells, ENCODE data) for the 500 kb region flanking eaQTLs. (c) Difference in raw H3K4me1 and H3K4me3 ChIP-seq signals for CAGE-peaks associated with puQTLs (promoters) and eaQTLs (enhancers). (d) CAGE signal at the *SWAP70* locus and associated enhancer region. The variant (rs12273601) mapped as puQTL for *SWAP70*-associated CAGE peak (p1) and eaQTL for paired enhancers (e1, e2) is shown. Normalized enhancer (e), promoter (f) and mRNA (g) expression relative to each genotype group are plotted for the entire population ( $n=154$ ).



**Supplementary Figure 8: assessing causal mediation of eaQTLs** for two model networks (**a**) considering either the enhancer activity as mediator for promoter expression (Model-1) or reciprocally (Model-2). (**b**) Distribution of Causal inference testing associated *p-values* for the model networks are plotted for the 158 triplets composed of eaQTLs being also puQTLs, associated enhancers and paired promoters. Significance threshold of *q-values* < 0.05 are shown (dashed lines). Number of tested trios in each significance group are indicated.



**Supplementary Table 1:** Samples sequencing information

| Cell Repository | Sample_ID | Sex    | Mapped_reads<br>(MAPQ 20) | Reads<br>redundancy |
|-----------------|-----------|--------|---------------------------|---------------------|
| Coriell         | GM06984   | male   | 12640183                  | 2.49                |
| Coriell         | GM06985   | female | 11969515                  | 2.26                |
| Coriell         | GM06989   | female | 20785454                  | 2.35                |
| Coriell         | GM07037   | female | 11086733                  | 2.4                 |
| Coriell         | GM07051   | male   | 14214959                  | 2.4                 |
| Coriell         | GM07056   | female | 9624480                   | 2.52                |
| Coriell         | GM07347   | male   | 16019204                  | 2.35                |
| Coriell         | GM07357   | male   | 15366979                  | 2.06                |
| Coriell         | GM10847   | female | 15738995                  | 2.48                |
| Coriell         | GM10851   | male   | 18737689                  | 2.15                |
| Coriell         | GM11829   | male   | 11997348                  | 2.3                 |
| Coriell         | GM11830   | female | 20129149                  | 2.46                |
| Coriell         | GM11831   | male   | 12089666                  | 2.23                |
| Coriell         | GM11832   | female | 13325612                  | 2.29                |
| Coriell         | GM11840   | female | 18607981                  | 2.13                |
| Coriell         | GM11843   | male   | 22446590                  | 2.33                |
| Coriell         | GM11881   | male   | 13457683                  | 2.52                |
| Coriell         | GM11893   | male   | 19222190                  | 2.45                |
| Coriell         | GM11894   | female | 10845659                  | 2.82                |
| Coriell         | GM11918   | female | 16428451                  | 2.75                |
| Coriell         | GM11919   | male   | 11620517                  | 2.61                |
| Coriell         | GM11920   | female | 15256307                  | 2.65                |
| Coriell         | GM11930   | male   | 12900853                  | 2.21                |
| Coriell         | GM11931   | female | 14685153                  | 2.35                |
| Coriell         | GM11932   | male   | 8830532                   | 2.5                 |
| Coriell         | GM11933   | female | 16240019                  | 2.53                |
| Coriell         | GM11992   | male   | 13437762                  | 2.29                |
| Coriell         | GM11994   | male   | 18510688                  | 2.27                |
| Coriell         | GM11995   | female | 16192878                  | 2.49                |
| Coriell         | GM12005   | male   | 16652720                  | 2.64                |
| Coriell         | GM12006   | female | 20833821                  | 2.49                |
| Coriell         | GM12043   | male   | 23134678                  | 2.32                |
| Coriell         | GM12044   | female | 15669075                  | 2.24                |
| Coriell         | GM12045   | male   | 16296316                  | 2.13                |
| Coriell         | GM12046   | female | 11436737                  | 2.35                |
| Coriell         | GM12154   | male   | 16587610                  | 2.75                |
| Coriell         | GM12155   | male   | 18328257                  | 2.48                |
| Coriell         | GM12156   | female | 15833961                  | 2.34                |
| Coriell         | GM12234   | female | 10459181                  | 2.12                |
| Coriell         | GM12249   | female | 18191955                  | 2.26                |
| Coriell         | GM12272   | male   | 18039600                  | 2.26                |
| Coriell         | GM12275   | female | 23622720                  | 2.83                |
| Coriell         | GM12282   | male   | 16212900                  | 2.42                |
| Coriell         | GM12283   | female | 19698012                  | 2.11                |
| Coriell         | GM12286   | male   | 11719353                  | 2.37                |
| Coriell         | GM12287   | female | 14583532                  | 2.33                |
| Coriell         | GM12340   | male   | 9413406                   | 1.98                |
| Coriell         | GM12341   | female | 14618466                  | 2.54                |
| Coriell         | GM12347   | male   | 15614874                  | 2.2                 |
| Coriell         | GM12348   | female | 10523527                  | 2.32                |
| Coriell         | GM12383   | female | 11837449                  | 2.35                |
| Coriell         | GM12399   | male   | 12531444                  | 2.52                |

|         |         |        |          |      |
|---------|---------|--------|----------|------|
| Coriell | GM12400 | female | 18129412 | 2.26 |
| Coriell | GM12413 | male   | 14593987 | 2.25 |
| Coriell | GM12414 | female | 14688668 | 2.34 |
| Coriell | GM12489 | female | 16032866 | 2.62 |
| Coriell | GM12546 | male   | 16380797 | 2.36 |
| Coriell | GM12716 | male   | 20138992 | 2.35 |
| Coriell | GM12717 | female | 6800531  | 2.38 |
| Coriell | GM12718 | female | 12489277 | 2.38 |
| Coriell | GM12748 | male   | 20231128 | 2.43 |
| Coriell | GM12749 | female | 16052947 | 2.62 |
| Coriell | GM12750 | male   | 20581013 | 2.36 |
| Coriell | GM12751 | female | 16631894 | 2.15 |
| Coriell | GM12760 | male   | 16089084 | 2.13 |
| Coriell | GM12761 | female | 16596991 | 2.06 |
| Coriell | GM12762 | male   | 14281839 | 2.19 |
| Coriell | GM12763 | female | 31132009 | 2.32 |
| Coriell | GM12775 | male   | 14567915 | 2.32 |
| Coriell | GM12776 | female | 16336452 | 2.12 |
| Coriell | GM12777 | male   | 12561614 | 1.95 |
| Coriell | GM12778 | female | 12540858 | 2.01 |
| Coriell | GM12813 | female | 11092562 | 2.2  |
| Coriell | GM12814 | male   | 21212043 | 2.51 |
| Coriell | GM12815 | female | 15694239 | 2.55 |
| Coriell | GM12827 | male   | 14480625 | 2.31 |
| Coriell | GM12828 | female | 12947084 | 2.36 |
| Coriell | GM12830 | female | 20600972 | 2.6  |
| Coriell | GM12842 | male   | 16326180 | 2.19 |
| Coriell | GM12843 | female | 16237630 | 2.06 |
| Coriell | GM12872 | male   | 13987532 | 2.22 |
| Coriell | GM12873 | female | 17346750 | 2.59 |
| Coriell | GM12874 | male   | 15848655 | 2.05 |
| Coriell | GM12878 | female | 14473727 | 2.47 |
| Coriell | GM12889 | male   | 20776334 | 2.29 |
| Coriell | GM12890 | female | 18365335 | 2.32 |
| GenCord | UCB1001 | female | 11247126 | 2.05 |
| GenCord | UCB1011 | male   | 6813096  | 2.2  |
| GenCord | UCB1012 | male   | 13519148 | 2.36 |
| GenCord | UCB1014 | female | 9561598  | 1.77 |
| GenCord | UCB1016 | female | 17149833 | 2.37 |
| GenCord | UCB1023 | female | 16187036 | 2.33 |
| GenCord | UCB1025 | male   | 16181650 | 2.53 |
| GenCord | UCB1027 | female | 6816136  | 1.81 |
| GenCord | UCB1029 | male   | 12970208 | 2.05 |
| GenCord | UCB1046 | male   | 15058736 | 2.34 |
| GenCord | UCB1048 | male   | 26478690 | 2.3  |
| GenCord | UCB1052 | female | 15574523 | 2.26 |
| GenCord | UCB1053 | female | 14323582 | 2.3  |
| GenCord | UCB1056 | female | 17783349 | 2.15 |
| GenCord | UCB1058 | female | 20343911 | 2.41 |
| GenCord | UCB1061 | male   | 18420911 | 2.33 |
| GenCord | UCB1064 | male   | 18428398 | 2.4  |
| GenCord | UCB1067 | male   | 18870632 | 2.31 |
| GenCord | UCB1069 | female | 17026777 | 2.09 |
| GenCord | UCB1070 | female | 13599139 | 2.31 |
| GenCord | UCB1071 | female | 20357006 | 2.61 |
| GenCord | UCB1072 | male   | 9735829  | 2.77 |
| GenCord | UCB1073 | male   | 14383787 | 2.7  |

|         |         |        |          |      |
|---------|---------|--------|----------|------|
| GenCord | UCB1074 | female | 20649343 | 2.43 |
| GenCord | UCB1075 | male   | 18363285 | 2.17 |
| GenCord | UCB1076 | female | 20469043 | 2.69 |
| GenCord | UCB1077 | male   | 22178879 | 2.72 |
| GenCord | UCB1079 | male   | 16944460 | 2    |
| GenCord | UCB1080 | male   | 16008585 | 2.21 |
| GenCord | UCB1083 | female | 20729535 | 2.18 |
| GenCord | UCB1086 | male   | 18305441 | 2.3  |
| GenCord | UCB1087 | male   | 20933257 | 2.14 |
| GenCord | UCB1089 | male   | 21579957 | 2.33 |
| GenCord | UCB1091 | female | 18696951 | 2.36 |
| GenCord | UCB1092 | female | 13986946 | 2.17 |
| GenCord | UCB1094 | female | 19336987 | 2.23 |
| GenCord | UCB1096 | male   | 27990927 | 2.7  |
| GenCord | UCB1098 | male   | 20290931 | 2.33 |
| GenCord | UCB1099 | female | 16722729 | 2.28 |
| GenCord | UCB1100 | male   | 14315030 | 2.69 |
| GenCord | UCB1101 | male   | 32925680 | 2.35 |
| GenCord | UCB1102 | female | 13679271 | 2.61 |
| GenCord | UCB1103 | male   | 15781322 | 2.78 |
| GenCord | UCB1105 | male   | 15540940 | 2.37 |
| GenCord | UCB1107 | male   | 19546188 | 2.26 |
| GenCord | UCB1108 | male   | 21124930 | 2.25 |
| GenCord | UCB1112 | male   | 15085524 | 2.61 |
| GenCord | UCB1113 | male   | 20592220 | 2.23 |
| GenCord | UCB1117 | female | 19965337 | 2.75 |
| GenCord | UCB1119 | female | 12269023 | 2.29 |
| GenCord | UCB1120 | male   | 22902656 | 2.61 |
| GenCord | UCB1129 | male   | 22146555 | 2.8  |
| GenCord | UCB1130 | male   | 11452870 | 2.37 |
| GenCord | UCB1131 | female | 11884848 | 1.79 |
| GenCord | UCB1136 | female | 24912880 | 2.73 |
| GenCord | UCB1139 | female | 10515970 | 2.04 |
| GenCord | UCB1148 | female | 10805484 | 2.49 |
| GenCord | UCB1196 | female | 12034373 | 2.74 |
| GenCord | UCB1199 | female | 12953368 | 2.02 |
| GenCord | UCB1200 | male   | 12520479 | 2.48 |
| GenCord | UCB1203 | male   | 13606302 | 1.78 |
| GenCord | UCB1206 | male   | 21348821 | 2.59 |
| GenCord | UCB1207 | female | 20464177 | 2.28 |
| GenCord | UCB1208 | male   | 13498234 | 2.22 |
| GenCord | UCB1210 | male   | 16064381 | 2.1  |
| GenCord | UCB1212 | male   | 15870193 | 2.18 |
| GenCord | UCB1216 | female | 17609259 | 2.77 |
| GenCord | UCB1218 | male   | 14569047 | 2.1  |

Supplementary Table 2: puQTLs linked with GWAS hits

| puQTLs (hg19, chromosome_start) | puQTLs associated promoter (hg19; chr_start_end_strand) | Promoter associated gene or regulatory element | GWAS hit (hg19, chromosome_start) | GWAS hit (rs ID) | RTC value | GWAS catalog reported trait(s) (reference PMID)                                                                           |
|---------------------------------|---------------------------------------------------------|------------------------------------------------|-----------------------------------|------------------|-----------|---------------------------------------------------------------------------------------------------------------------------|
| chr8_11338383                   | chr8_11324375_11324387_-                                | <i>FAM167A</i> (ENSG00000154319.10)            | chr8_11339965                     | rs2736332        | 1         | systemic lupus erythematosus (PMID:27399966;26502338)                                                                     |
| chr8_11339965                   | chr8_11324273_11324289_-                                | <i>FAM167A</i> (ENSG00000154319.10)            | chr8_11339965                     | rs2736332        | 1         | systemic lupus erythematosus (PMID:27399966;26502338)                                                                     |
| chr8_11351019                   | chr8_11313812_11313825_-                                | intronic enhancer element                      | chr8_11339965                     | rs2736332        | 1         | systemic lupus erythematosus (PMID:27399966;26502338)                                                                     |
| chr8_11351912                   | chr8_11313523_11313532_-                                | intronic ( <i>FAM167A</i> )                    | chr8_11339965                     | rs2736332        | 1         | systemic lupus erythematosus (PMID:27399966;26502338)                                                                     |
| chr8_11352541                   | chr8_11313428_11313437_-                                | intronic ( <i>FAM167A</i> )                    | chr8_11352541                     | rs2618476        | 1         | systemic lupus erythematosus (PMID:19165918)                                                                              |
| chr8_11352541                   | chr8_11313428_11313437_-                                | intronic ( <i>FAM167A</i> )                    | chr8_11339965                     | rs2736332        | 0.923     | systemic lupus erythematosus (PMID:27399966;26502338)                                                                     |
| chr8_11395232                   | chr8_11351905_11351925_+                                | <i>BLK</i> (ENSG00000136573.8)                 | chr8_11395232                     | rs1478897        | 1         | systemic lupus erythematosus (PMID:26316170)                                                                              |
| chr8_11403769                   | chr8_11403533_11403544_+                                | <i>BLK</i> (ENSG00000136573.8)                 | chr8_11339965                     | rs2736332        | 1         | systemic lupus erythematosus (PMID:27399966;26502338)                                                                     |
| chr8_11453671                   | chr8_11665932_11665943_+                                | <i>FDFIT1</i> (ENSG00000079459.8)              | chr8_11352541                     | rs2618476        | 0.923     | systemic lupus erythematosus (PMID:19165918)                                                                              |
| chr1_67731368                   | chr1_67519812_67519854_-                                | <i>SLC35D1</i> (ENSG00000116704.6)             | chr1_67731368                     | rs7547569        | 1         | inflammatory bowel disease (PMID:26192919)                                                                                |
| chr13_44453480                  | chr13_44453833_44453912_-                               | <i>CCDC122</i> (ENSG00000151773.8)             | chr13_44457925                    | rs3764147        | 1         | inflammatory bowel disease (PMID:26192919); Crohn's disease (PMID:23128233;21102463;18587394); leprosy (PMID:20018961)    |
| chr13_44457925                  | chr13_44453688_44453795_+                               | <i>LACC1</i> (ENSG00000179630.6)               | chr13_44457925                    | rs3764147        | 1         | inflammatory bowel disease (PMID:26192919); Crohn's disease (PMID:23128233;21102463;18587394); leprosy (PMID:20018961)    |
| chr16_28837515                  | chr16_28857630_28857675_+                               | <i>TUFM</i> (ENSG00000178952.4)                | chr16_28837515                    | rs8049439        | 1         | inflammatory bowel disease (PMID:19915574); educational attainment (PMID:27225129;25201988;23722424)                      |
| chr19_10512400                  | chr19_10515518_10515566_+                               | intronic enhancer element                      | chr19_10512911                    | rs11879191       | 1         | inflammatory bowel disease (PMID:23128233); Crohn's disease (PMID:26192919)                                               |
| chr19_10512911                  | chr19_10515604_10515629_+                               | intronic enhancer element                      | chr19_10512911                    | rs11879191       | 1         | inflammatory bowel disease (PMID:23128233); Crohn's disease (PMID:26192919)                                               |
| chr9_4985879                    | chr9_4985228_4985245_+                                  | <i>JAK2</i> (ENSG00000096968.8)                | chr9_4985879                      | rs2274471        | 1         | Crohn's disease (PMID:23266558)                                                                                           |
| chr19_40170053                  | chr19_40170009_40170016_+                               | <i>LGALS17A</i> (ENSG00000226025.5)            | chr19_40170053                    | rs8103033        | 1         | obesity-related traits (PMID:23251661)                                                                                    |
| chr22_42326181                  | chr22_42342702_42342716_-                               | <i>CENPM</i> (ENSG00000100162.10)              | chr22_42336172                    | rs5758511        | 1         | birth weight (PMID:23202124)                                                                                              |
| chr22_42336172                  | chr22_42336209_42336228_-                               | <i>CENPM</i> (ENSG00000100162.10)              | chr22_42336172                    | rs5758511        | 1         | birth weight (PMID:23202124)                                                                                              |
| chr3_16413520                   | chr3_16524357_16524377_-                                | <i>RFTN1</i> (ENSG00000131378.9)               | chr3_16417555                     | rs493733         | 1         | urate levels, BMI interaction (PMID:25811787)                                                                             |
| chr3_16417555                   | chr3_16553570_16553583_+                                | antisense transcript of <i>RFTN1</i>           | chr3_16417555                     | rs493733         | 1         | urate levels, BMI interaction (PMID:25811787)                                                                             |
| chr3_52648265                   | chr3_52719912_52719995_-                                | <i>PBRM1</i> (ENSG00000163939.14)              | chr3_52649748                     | rs13083798       | 1         | waist-hip ratio adjusted for body mass index (PMID:25673412)                                                              |
| chr3_52649748                   | chr3_52720016_52720057_+                                | <i>GNL3</i> (ENSG00000163938.12)               | chr3_52649748                     | rs13083798       | 1         | waist-hip ratio adjusted for body mass index (PMID:25673412)                                                              |
| chr1_11862778                   | chr1_11866270_11866314_+                                | <i>CLCN6</i> (ENSG0000011021.17)               | chr1_11862778                     | rs17367504       | 1         | blood pressure (PMID:21909110); systolic blood pressure (PMID:19430483)                                                   |
| chr1_89467242                   | chr1_89458607_89458627_-                                | <i>CCBL2</i> (ENSG00000213516.5)               | chr1_89474818                     | rs17433780       | 1         | carotid artery intima media thickness (PMID:26343869)                                                                     |
| chr1_89474818                   | chr1_89458651_89458685_-                                | <i>CCBL2</i> (ENSG00000213516.5)               | chr1_89474818                     | rs17433780       | 1         | carotid artery intima media thickness (PMID:26343869)                                                                     |
| chr1_89618472                   | chr1_89458415_89458463_-                                | <i>CCBL2</i> (ENSG00000213516.5)               | chr1_89474818                     | rs17433780       | 1         | carotid artery intima media thickness (PMID:26343869)                                                                     |
| chr2_203808532                  | chr2_203736475_203736503_-                              | <i>ICA1L</i> (ENSG00000163596.12)              | chr2_203808532                    | rs140244541      | 1         | low density lipoprotein cholesterol measurement (PMID:25961943)                                                           |
| chr2_85788175                   | chr2_85788605_85788629_-                                | <i>GGCX</i> (ENSG00000115486.7)                | chr2_85788175                     | rs1568458        | 1         | coronary artery disease (PMID:26343387)                                                                                   |
| chr15_78817929                  | chr15_78832757_78832806_+                               | <i>PSMA4</i> (ENSG00000041357.11)              | chr15_78817929                    | rs8042849        | 1         | forced expiratory volume post bronchodilator (PMID:26634245)                                                              |
| chr15_78817929                  | chr15_78832757_78832806_+                               | <i>PSMA4</i> (ENSG00000041357.11)              | chr15_78821016                    | rs12441354       | 1         | forced expiratory volume post bronchodilator (PMID:26634245)                                                              |
| chr15_78821016                  | chr15_78832993_78833010_-                               | antisense transcript of <i>PSMA4</i>           | chr15_78821016                    | rs12441354       | 1         | forced expiratory volume post bronchodilator (PMID:26634245)                                                              |
| chr15_78857812                  | chr15_78857836_78857847_+                               | <i>CHRNA5</i> (ENSG00000169684.9)              | chr15_78817929                    | rs8042849        | 1         | forced expiratory volume post bronchodilator (PMID:26634245)                                                              |
| chr15_78858400                  | chr15_78857870_78857897_+                               | <i>CHRNA5</i> (ENSG00000169684.9)              | chr15_78858400                    | rs684513         | 1         | forced expiratory volume post bronchodilator (PMID:26634245)                                                              |
| chr1_155194980                  | chr1_155197244_155197268_-                              | <i>GBAP1</i> (ENSG00000160766.10)              | chr1_155194980                    | rs2049805        | 1         | renal function related traits (PMID:22797727)                                                                             |
| chr11_44087989                  | chr11_44088027_44088086_+                               | <i>ACCS</i> (ENSG00000110455.9)                | chr11_44087989                    | rs2074038        | 1         | IGA glomerulonephritis (PMID:26028593)                                                                                    |
| chr20_23612737                  | chr20_23618615_23618627_-                               | <i>CST3</i> (ENSG00000101439.4)                | chr20_23612737                    | rs911119         | 1         | chronic kidney disease (PMID:20383146)                                                                                    |
| chr11_61595564                  | chr11_61595533_61595544_+                               | <i>FADS2</i> (ENSG00000134824.9)               | chr11_61595564                    | rs968567         | 1         | rheumatoid arthritis (PMID:24390342); glycerophospholipid levels (PMID:26068415); blood metabolite levels (PMID:24816252) |
| chr22_39747671                  | chr22_39745939_39746012_+                               | <i>SYNGR1</i> (ENSG00000100321.10)             | chr22_39747671                    | rs909685         | 1         | rheumatoid arthritis (PMID:24390342)                                                                                      |
| chr13_76120727                  | chr13_76123613_76123659_+                               | <i>UCHL3</i> (ENSG00000118939.13)              | chr13_76136648                    | rs9543976        | 1         | diabetic retinopathy (PMID:23562823)                                                                                      |
| chr13_76136648                  | chr13_76123883_76123949_+                               | <i>UCHL3</i> (ENSG00000118939.13)              | chr13_76136648                    | rs9543976        | 1         | diabetic retinopathy (PMID:23562823)                                                                                      |
| chr12_56401085                  | chr12_56435881_56435936_+                               | <i>RPS26</i> (ENSG00000197728.5)               | chr12_56401085                    | rs10876864       | 1         | viteligo (PMID:22951725)                                                                                                  |
| chr12_56466654                  | chr12_56122977_56123016_+                               | <i>CD63</i> (ENSG00000135404.7)                | chr12_56401085                    | rs10876864       | 1         | viteligo (PMID:22951725)                                                                                                  |
| chr1_150826831                  | chr1_150947192_150947209_-                              | <i>CERS2</i> (ENSG00000143418.15)              | chr1_150860471                    | rs7412746        | 1         | melanoma (PMID:21983785)                                                                                                  |
| chr1_150860471                  | chr1_150738211_150738260_-                              | <i>CST5</i> (ENSG00000163131.6)                | chr1_150860471                    | rs7412746        | 1         | melanoma (PMID:21983785)                                                                                                  |
| chr10_131265545                 | chr10_131265443_131265495_+                             | <i>MGMT</i> (ENSG00000170430.9)                | chr10_131265545                   | rs16906252       | 1         | <i>MGMT</i> methylation in smokers (PMID:26183928)                                                                        |
| chr14_24609147                  | chr14_24610694_24610724_-                               | <i>FAM158A</i> (ENSG00000100908.9)             | chr14_24609147                    | rs8010715        | 1         | serum IgG glycosylation measurement (PMID:23382691)                                                                       |
| chr16_792190                    | chr16_777097_777112_+                                   | <i>HAGHL</i> (ENSG00000103253.13)              | chr16_792190                      | rs11648796       | 1         | body height (PMID:25429064;20881960)                                                                                      |
| chr19_2214057                   | chr19_2236246_2236286_-                                 | <i>PLEKHJ1</i> (ENSG00000104886.5)             | chr19_2214057                     | rs2074552        | 1         | plasma omega-6 polyunsaturated fatty acid levels (PMID:26584805)                                                          |
| chr2_208012509                  | chr2_208031064_208031075_+                              | antisense transcript of <i>KLF7</i>            | chr2_208012509                    | rs2284932        | 1         | lacrimal and salivary gland lesion in type 1 autoimmune pancreatitis (PMID:25985088)                                      |
| chr5_125918148                  | chr5_125930877_125930919_-                              | <i>ALDH7A1</i> (ENSG00000164904.11)            | chr5_125918148                    | rs13182402       | 1         | osteoporosis (PMID:20072603)                                                                                              |
| chr6_147531081                  | chr6_147524975_147524988_+                              | <i>STXBPS</i> (ENSG00000164506.10)             | chr6_147531081                    | rs2786189        | 1         | bronchopulmonary dysplasia (PMID:23897914)                                                                                |
| chr6_32604372                   | chr6_32610846_32610863_+                                | <i>HLA-DQA1</i> (ENSG00000196735.7)            | chr6_32604372                     | rs9272346        | 1         | asthma (PMID:23181788); type 1 diabetes mellitus (PMID:18978792;17554300)                                                 |

**Supplementary Table 3:**  
Mapped reads of GEUVADIS  
RNA-seq samples

| <b>Sample_ID</b> | <b>Mapped_reads</b> |
|------------------|---------------------|
| GM06984          | 2.39E+07            |
| GM06985          | 4.27E+07            |
| GM06986          | 4.51E+07            |
| GM06989          | 2.66E+07            |
| GM06994          | 4.74E+07            |
| GM07037          | 3.54E+07            |
| GM07048          | 3.24E+07            |
| GM07051          | 3.47E+07            |
| GM07056          | 3.24E+07            |
| GM07347          | 4.28E+07            |
| GM07357          | 3.32E+07            |
| GM10847          | 4.05E+07            |
| GM10851          | 3.53E+07            |
| GM11829          | 2.71E+07            |
| GM11830          | 3.25E+07            |
| GM11831          | 5.35E+07            |
| GM11832          | 4.62E+07            |
| GM11840          | 3.46E+07            |
| GM11843          | 3.80E+07            |
| GM11881          | 3.85E+07            |
| GM11892          | 2.76E+07            |
| GM11893          | 5.39E+07            |
| GM11894          | 4.59E+07            |
| GM11918          | 3.12E+07            |
| GM11920          | 4.16E+07            |
| GM11930          | 2.68E+07            |
| GM11931          | 3.29E+07            |
| GM11992          | 2.99E+07            |
| GM11994          | 5.85E+07            |
| GM11995          | 3.41E+07            |
| GM12004          | 2.52E+07            |
| GM12005          | 3.97E+07            |
| GM12006          | 4.38E+07            |
| GM12043          | 4.15E+07            |
| GM12044          | 4.12E+07            |
| GM12045          | 2.01E+07            |
| GM12058          | 4.07E+07            |
| GM12144          | 4.06E+07            |
| GM12154          | 3.59E+07            |

|         |          |
|---------|----------|
| GM12155 | 3.19E+07 |
| GM12156 | 4.02E+07 |
| GM12234 | 4.44E+07 |
| GM12249 | 3.55E+07 |
| GM12272 | 2.31E+07 |
| GM12273 | 3.41E+07 |
| GM12275 | 3.83E+07 |
| GM12282 | 2.02E+07 |
| GM12283 | 3.22E+07 |
| GM12286 | 3.87E+07 |
| GM12287 | 1.87E+07 |
| GM12340 | 3.94E+07 |
| GM12341 | 3.86E+07 |
| GM12342 | 3.45E+07 |
| GM12347 | 3.85E+07 |
| GM12348 | 1.92E+07 |
| GM12383 | 3.21E+07 |
| GM12399 | 1.53E+07 |
| GM12400 | 3.30E+07 |
| GM12413 | 4.99E+07 |
| GM12489 | 3.81E+07 |
| GM12546 | 2.54E+07 |
| GM12716 | 9.19E+07 |
| GM12717 | 3.55E+07 |
| GM12718 | 4.39E+07 |
| GM12749 | 2.64E+07 |
| GM12750 | 3.63E+07 |
| GM12751 | 4.90E+07 |
| GM12760 | 1.63E+07 |
| GM12761 | 4.18E+07 |
| GM12762 | 2.39E+07 |
| GM12763 | 4.29E+07 |
| GM12775 | 5.73E+07 |
| GM12776 | 4.17E+07 |
| GM12777 | 3.52E+07 |
| GM12778 | 3.20E+07 |
| GM12812 | 2.68E+07 |
| GM12813 | 3.48E+07 |
| GM12814 | 5.71E+07 |
| GM12815 | 3.97E+07 |
| GM12827 | 1.98E+07 |
| GM12829 | 5.36E+07 |
| GM12830 | 2.45E+07 |

|         |          |         |          |
|---------|----------|---------|----------|
| GM12842 | 4.41E+07 | GM20536 | 2.99E+07 |
| GM12843 | 2.99E+07 | GM20538 | 2.06E+07 |
| GM12872 | 4.30E+07 | GM20539 | 3.77E+07 |
| GM12873 | 3.92E+07 | GM20540 | 2.47E+07 |
| GM12874 | 3.62E+07 | GM20541 | 4.03E+07 |
| GM12889 | 4.37E+07 | GM20542 | 2.36E+07 |
| GM12890 | 3.08E+07 | GM20543 | 3.60E+07 |
| GM20502 | 3.22E+07 | GM20544 | 3.70E+07 |
| GM20503 | 2.88E+07 | GM20581 | 2.93E+07 |
| GM20504 | 3.53E+07 | GM20582 | 4.46E+07 |
| GM20505 | 3.40E+07 | GM20585 | 2.64E+07 |
| GM20506 | 2.91E+07 | GM20586 | 4.29E+07 |
| GM20507 | 3.47E+07 | GM20588 | 4.03E+07 |
| GM20508 | 3.73E+07 | GM20589 | 3.57E+07 |
| GM20509 | 7.60E+07 | GM20752 | 4.75E+07 |
| GM20510 | 1.74E+07 | GM20754 | 3.55E+07 |
| GM20512 | 3.46E+07 | GM20756 | 3.77E+07 |
| GM20513 | 3.88E+07 | GM20757 | 3.81E+07 |
| GM20514 | 2.99E+07 | GM20758 | 5.59E+07 |
| GM20515 | 4.81E+07 | GM20759 | 2.30E+07 |
| GM20516 | 4.05E+07 | GM20760 | 2.36E+07 |
| GM20517 | 2.71E+07 | GM20761 | 3.61E+07 |
| GM20518 | 3.14E+07 | GM20765 | 5.50E+07 |
| GM20519 | 4.15E+07 | GM20766 | 3.26E+07 |
| GM20520 | 1.82E+07 | GM20768 | 3.68E+07 |
| GM20521 | 3.04E+07 | GM20769 | 2.80E+07 |
| GM20524 | 4.40E+07 | GM20770 | 2.01E+07 |
| GM20525 | 2.97E+07 | GM20771 | 4.00E+07 |
| GM20527 | 5.29E+07 | GM20772 | 3.82E+07 |
| GM20528 | 4.14E+07 | GM20773 | 3.33E+07 |
| GM20529 | 4.01E+07 | GM20774 | 2.40E+07 |
| GM20530 | 4.74E+07 | GM20778 | 1.25E+07 |
| GM20531 | 3.97E+07 | GM20783 | 4.27E+07 |
| GM20532 | 2.36E+07 | GM20785 | 3.46E+07 |
| GM20534 | 4.54E+07 | GM20786 | 4.53E+07 |
| GM20535 | 3.30E+07 | GM20787 | 4.34E+07 |