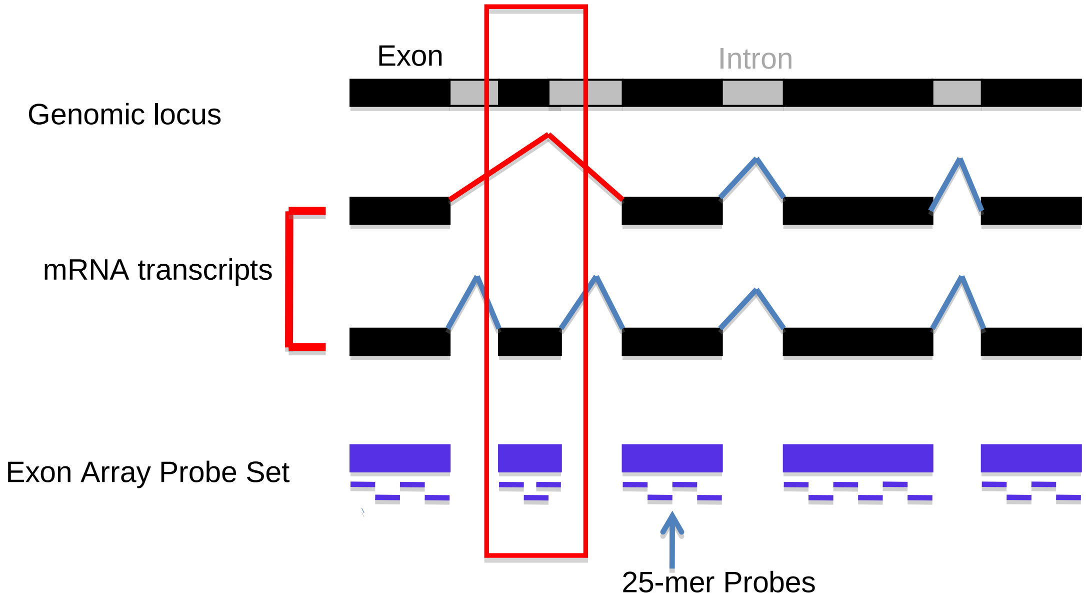
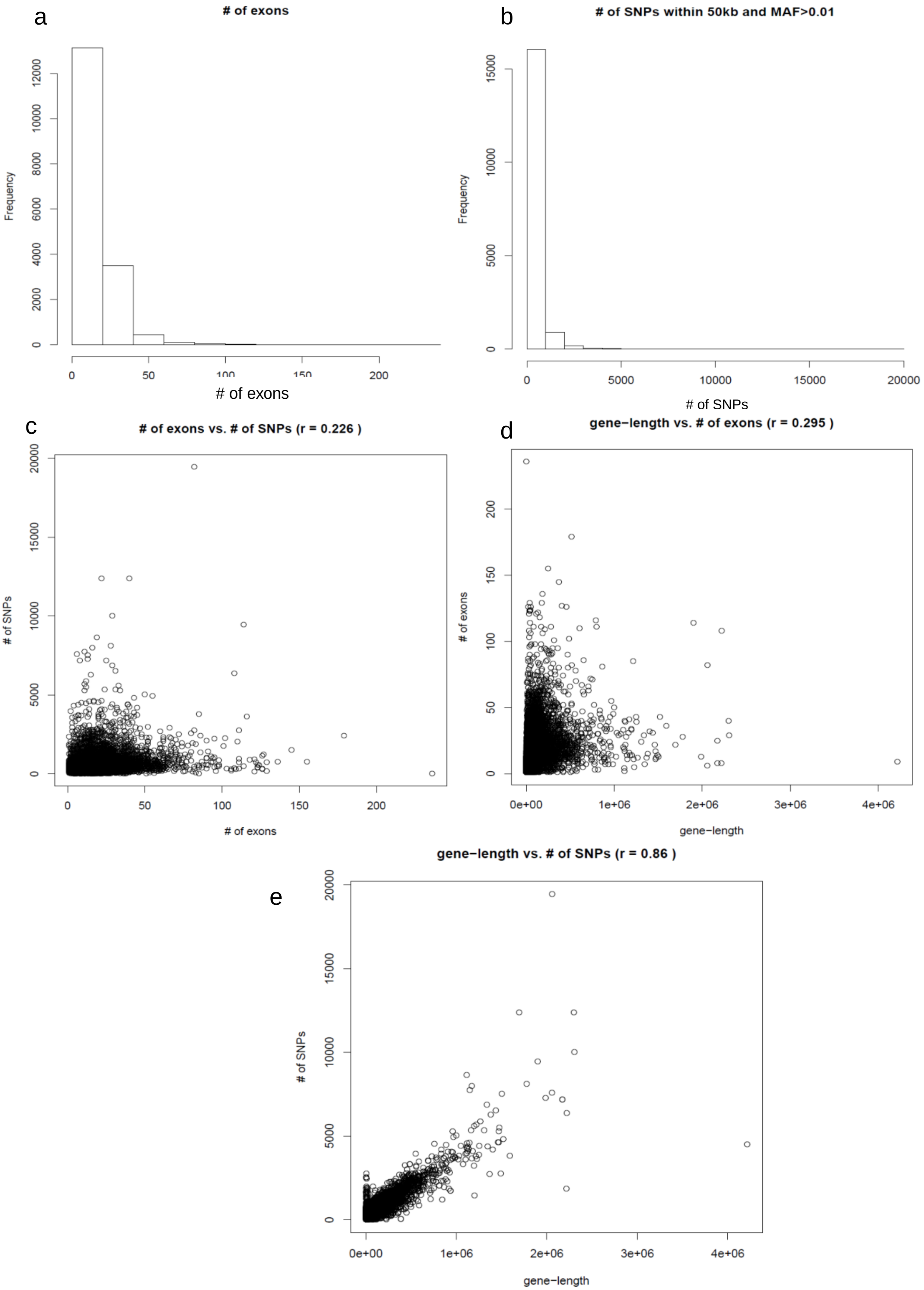


Supplementary Figure 1

Schematic for coverage of Affymetrix exon array probe sets across the entire length of the transcript.

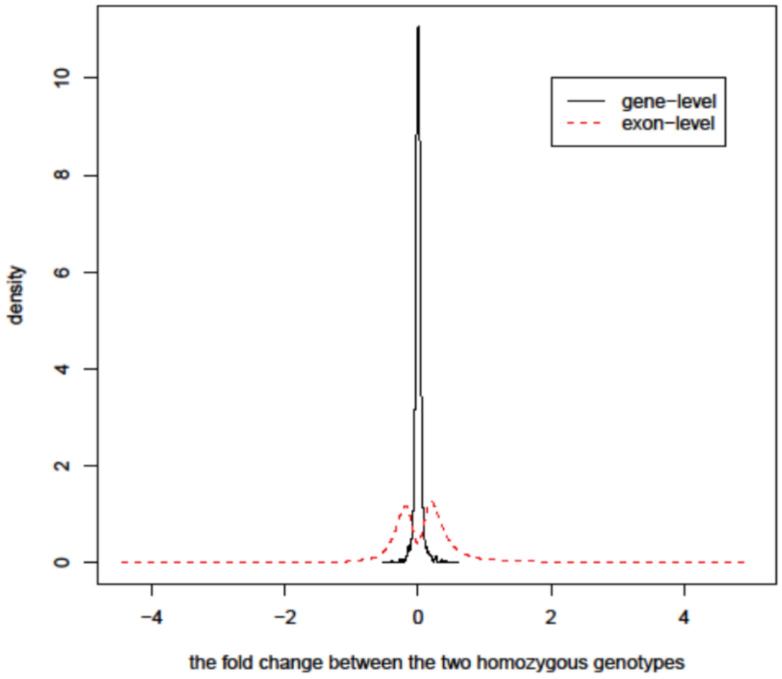
Black regions represent exons, whereas gray regions represent introns. The short dashes underneath the exon regions indicate individual probes of 25 nt in length representing the probe set. The highlighted red box indicates an alternative splicing event (exon 2 is spliced out in mRNA transcript isoform 1), which can be detected by analyzing the exon-level probe sets one by one in this genomic locus. The Affymetrix GeneChip Human Exon 1.0 ST array allows for exon-level expression profiling on a single chip and can interrogate over 280,000 core exons in the human genome.


a
# of exons
Frequency
0
2000
4000
6000
8000
10000
12000
0
50
100
150
200
# of exons
b
# of SNPs within 50kb and MAF>0.01
Frequency
0
5000
10000
15000
0
5000
10000
15000
20000
# of SNPs
c
# of exons vs. # of SNPs (r = 0.226)
# of SNPs
0
5000
10000
15000
20000
0
50
100
150
200
# of exons
d
gene−length vs. # of exons (r = 0.295)
# of exons
0
50
100
150
200
0e+00
1e+06
2e+06
3e+06
4e+06
gene−length
e
gene−length vs. # of SNPs (r = 0.86)
# of SNPs
0
5000
10000
15000
20000
0e+00
1e+06
2e+06
3e+06
4e+06
gene−length

Supplementary Figure 2

The number of exons and SNPs versus the gene length.

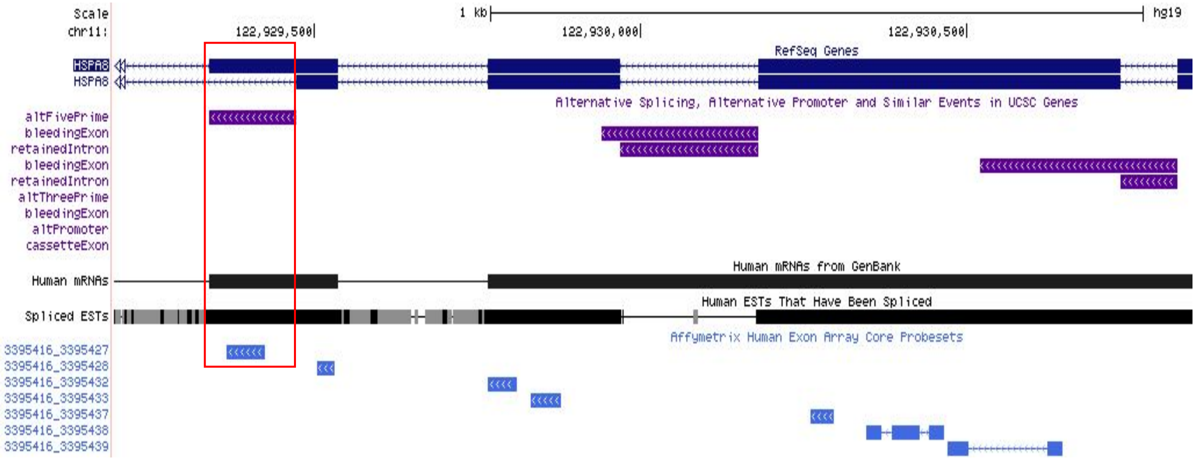
Histograms of (**a**) the number of core probe sets/exons per gene and (**b**) the number of SNPs with minor allele frequency (MAF) > 0.01 within 50 kb of each gene. Correlation plots of (**c**) the number of probe sets/exons to the number of SNPs located within 50 kb of each gene, (**d**) the length of genes to the number of probe sets/exons and (**e**) the number of SNPs located within 50 kb of each gene.



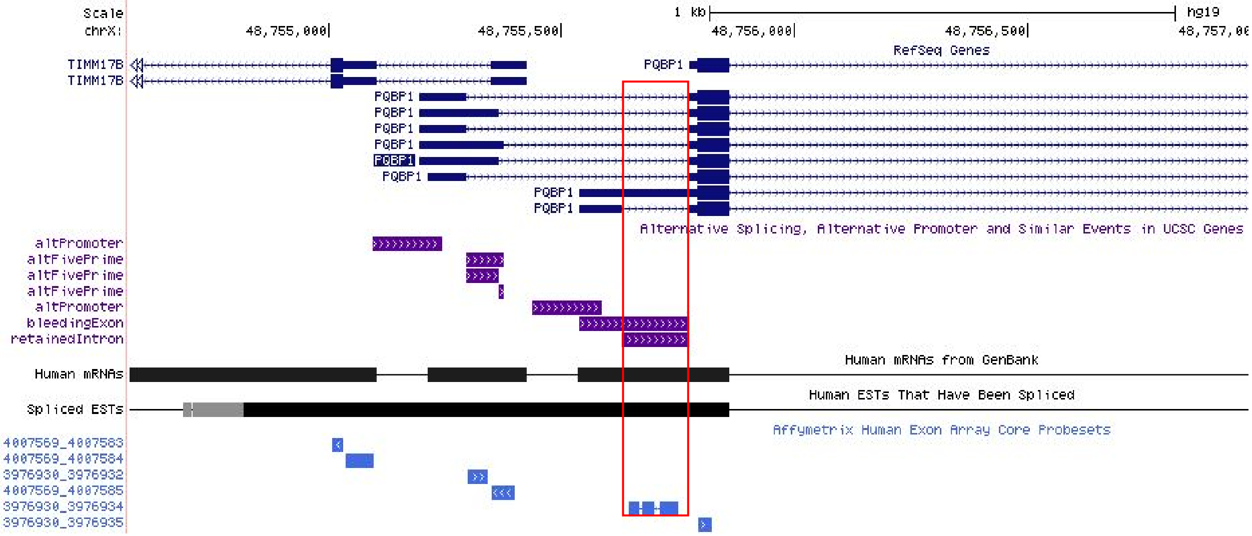
Supplementary Figure 3

For the 2,650 genes only found in exon-level analysis, shown is the distribution of fold differences in expression levels between the 2 homozygous genotypes.

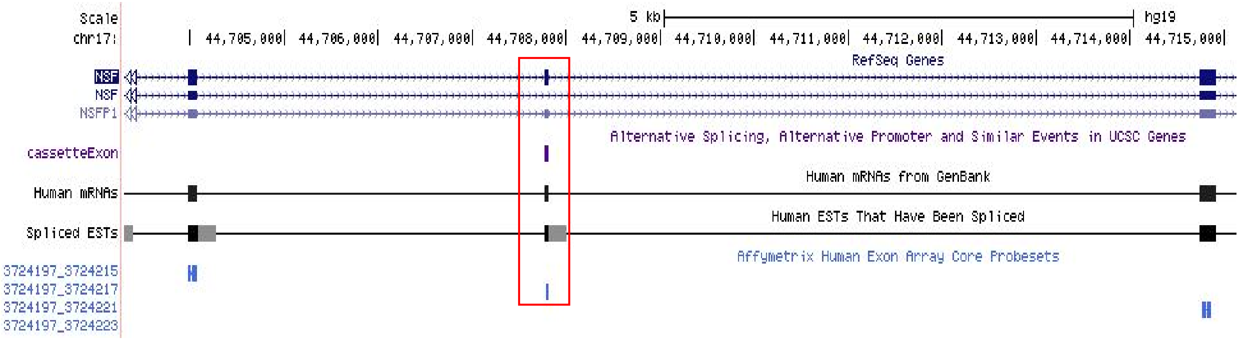
a. Alternate 5’ End: *HSPA8*, PS 3395427 vs. rs1064585



b. Retained Intron: *PQBP1*, PS 3976934 vs. rs112261029



c. Cassette Exon: *NSF*, PS 3724217 vs. rs199532

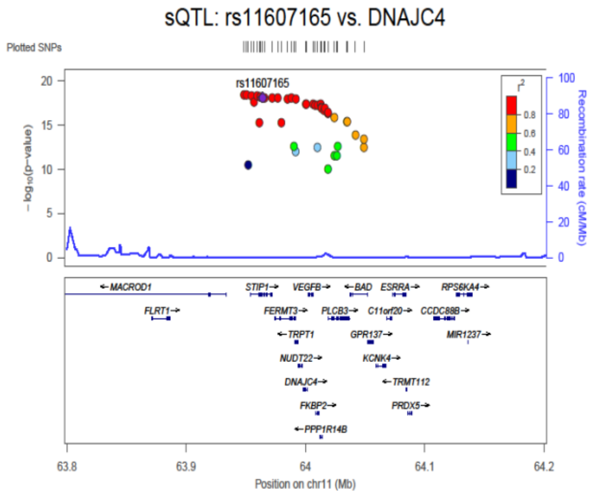


Supplementary Figure 4

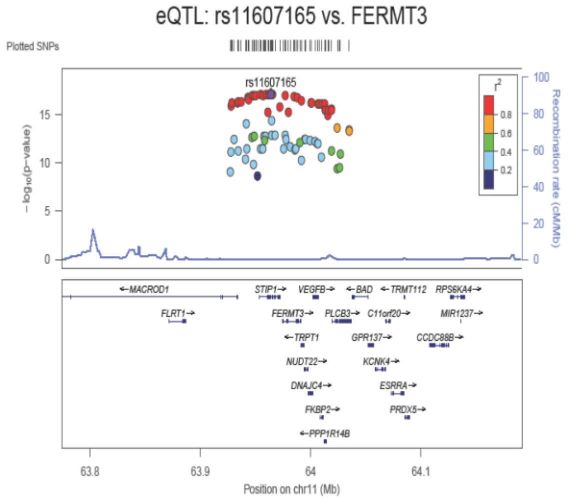
Examples of the different types of transcript isoform events observed.

a. The GWAS SNP rs11607165 on chr11

1. The index SNP rs11607165 is the top ***cis*-sQTL** for gene *DNAJC4*

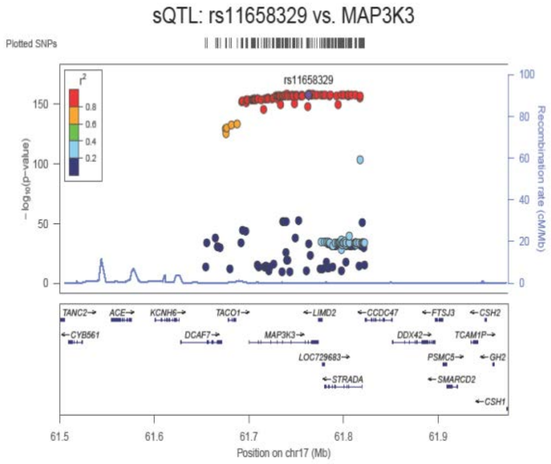

sQTL: rs11607165 vs. DNAJC4

2. The index SNP rs11607165 is the top ***cis*-eQTL** for gene *FERMT3*

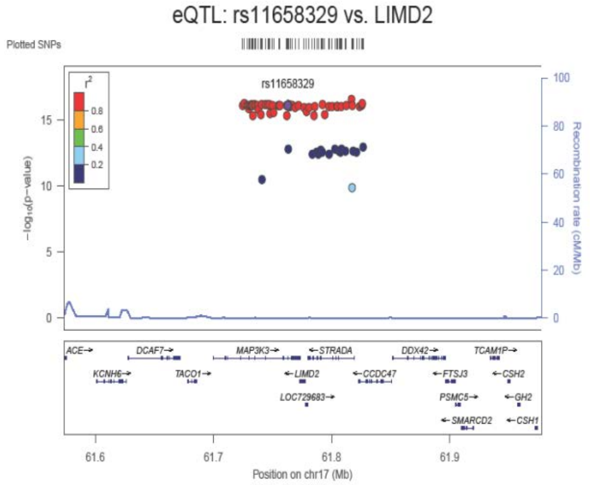

eQTL: rs11607165 vs. FERMT3

b. The GWAS SNP rs11658329 on chr17

1. The index SNP rs11658329 is the top ***cis*-sQTL** for gene *MAP3K3*

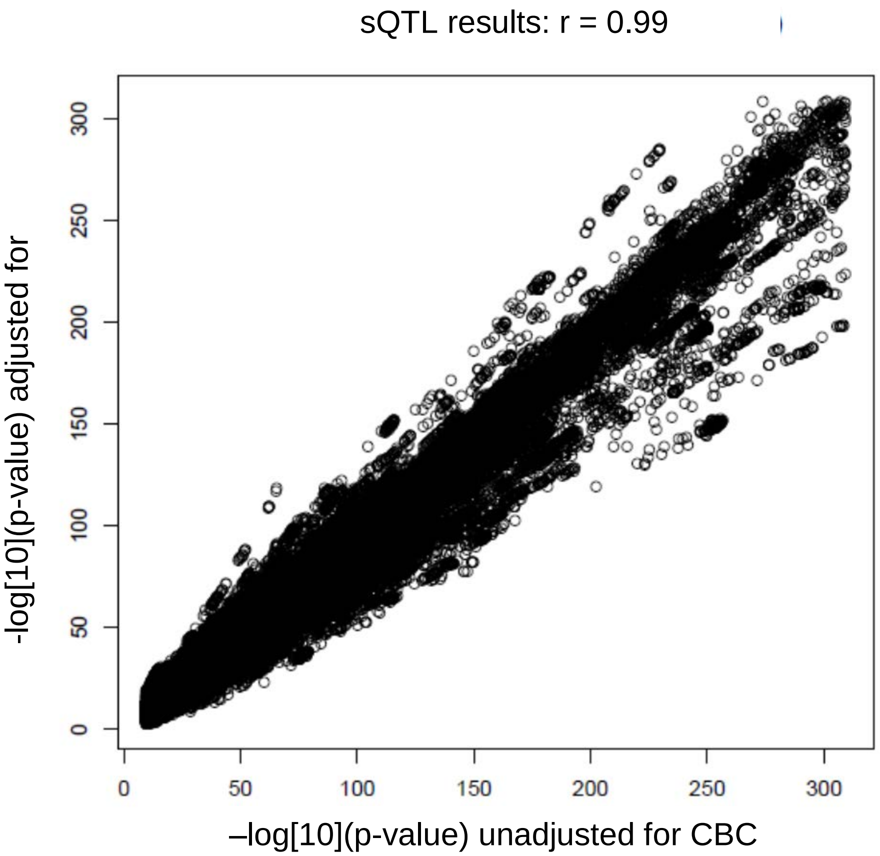

sQTL: rs11658329 vs. MAP3K3

2. The index SNP rs11658329 is the top ***cis*-eQTL** for gene *LIMD2*


eQTL: rs11658329 vs. LIMD2

Supplementary Figure 5

Examples of two GWAS SNPs that are in close LD ($r^2 > 0.8$) with both the peak signal of a *cis* eQTL and the peak signal of a *cis* sQTL (the index SNP is represented as a purple diamond).



Supplementary Figure 6

The correlation plot of the –log(*P* values) of *cis*-sQTL results with and without adjusting for the cell counts of seven major blood cell types: total white blood cell count (WBC), total platelet count (PLT), and subfractions (percentages) of neutrophils, lymphocytes, monocytes, eosinophils and basophils.