The American Journal of Human Genetics, *92*

# Supplemental Data

# Integrating GWAS and Expression Data for Functional Characterization of Disease-Associated SNPs: An Application to Follicular Lymphoma

Lucia Conde, Paige M. Bracci, Rhea Richardson, Stephen B. Montgomery, and Christine F. Skibola

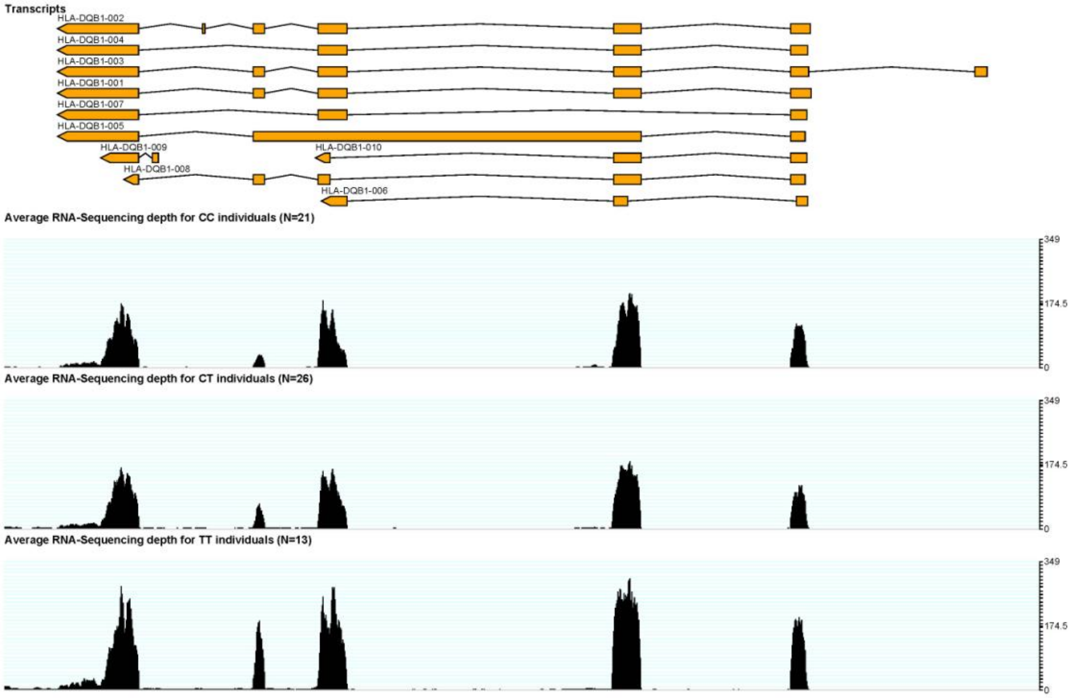

**Figure S1**. **Average HLA-DQB1 Sequencing Depth in the E-MTAB-197 RNA-Seq Data Set Stratified by rs2647012 Genotype**

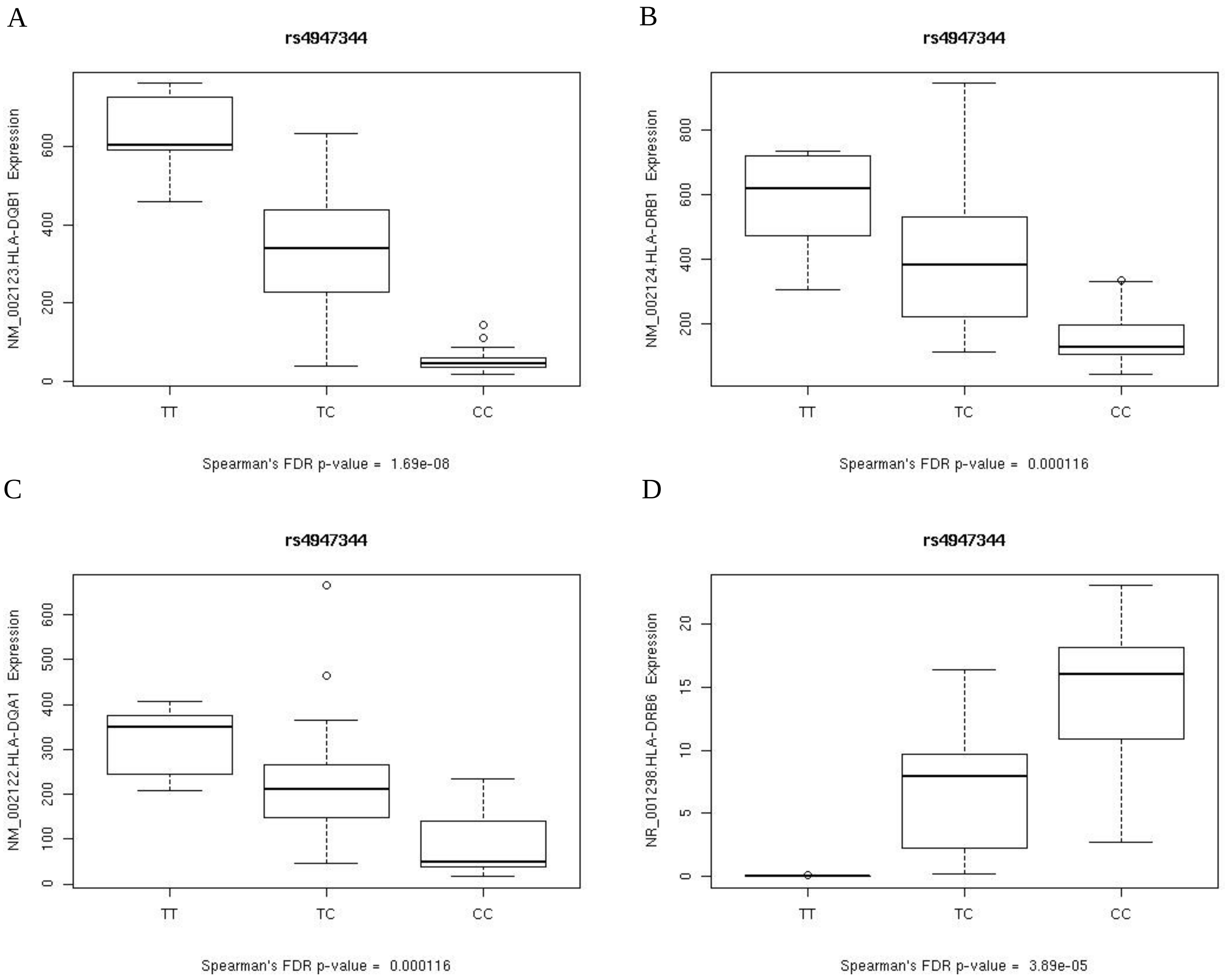

**Figure S2**. **Correlation of rs4947344 with (A)** ***HLA-DQB1*****, (B)** ***HLA-DRB1*****, (C)** ***HLA-DQA1*****, and (D)** ***HLA-DRB6*** **Expression Levels in the GSE16921 RNA-Seq Data Set**

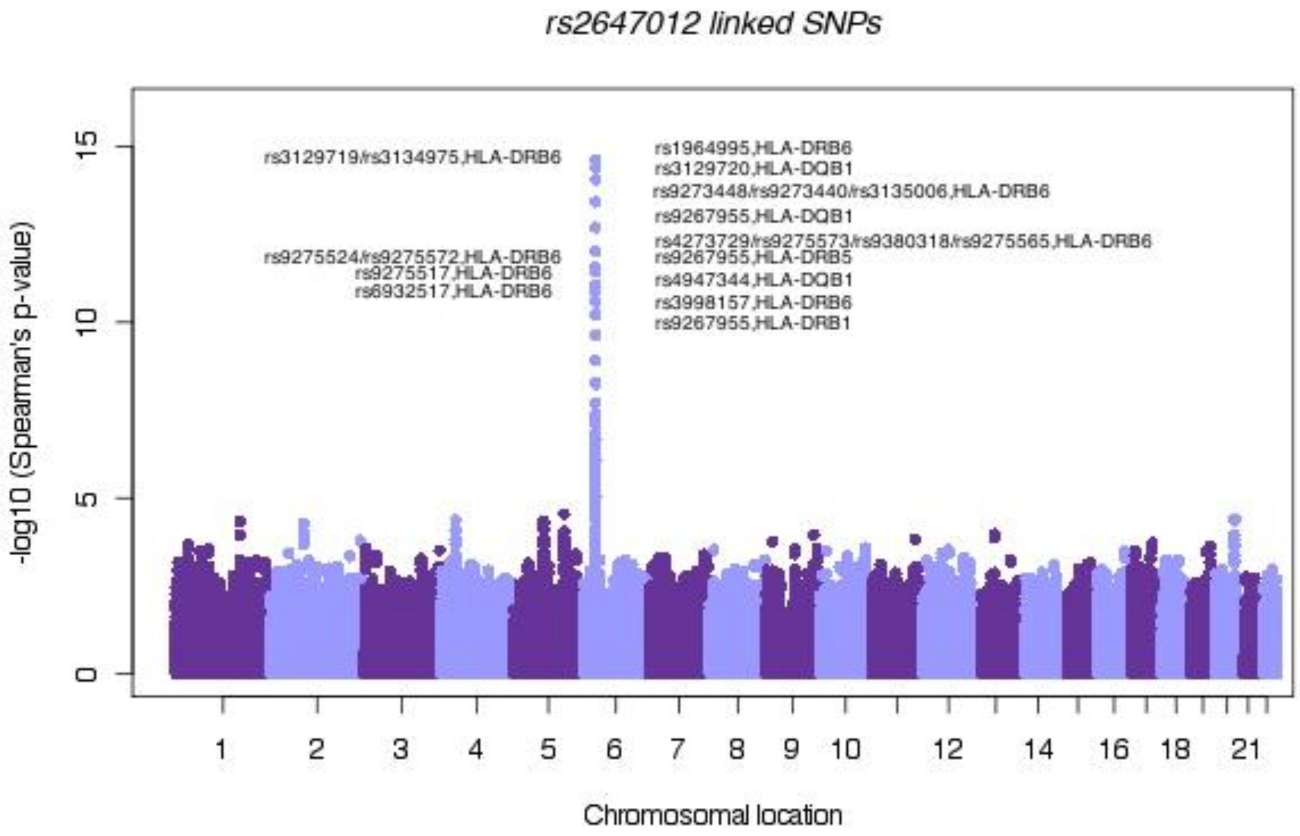

**Figure S3**. **Genome-wide SNP-Probe Associations for rs2647012-Linked SNPs**

The strongest associations, represented as $-\log_{10}$ of the Spearman's observed p-value, are observed for HLA class II genes located in cis with respect to the rs2647012-linked SNPs. Association p-values were obtained using the GSE16921 dataset.

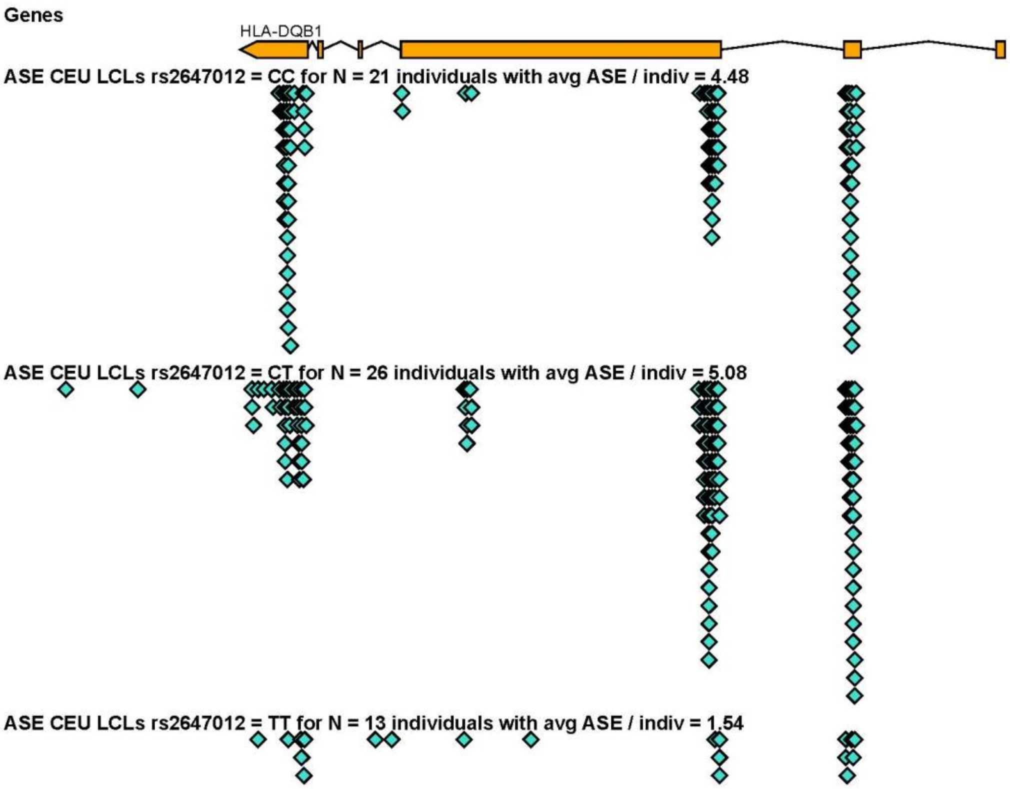

**Figure S4**. **Representation of the Total Number of Significant *HLA-DQB1* ASE Sites Stratified by rs2647012 Genotype**

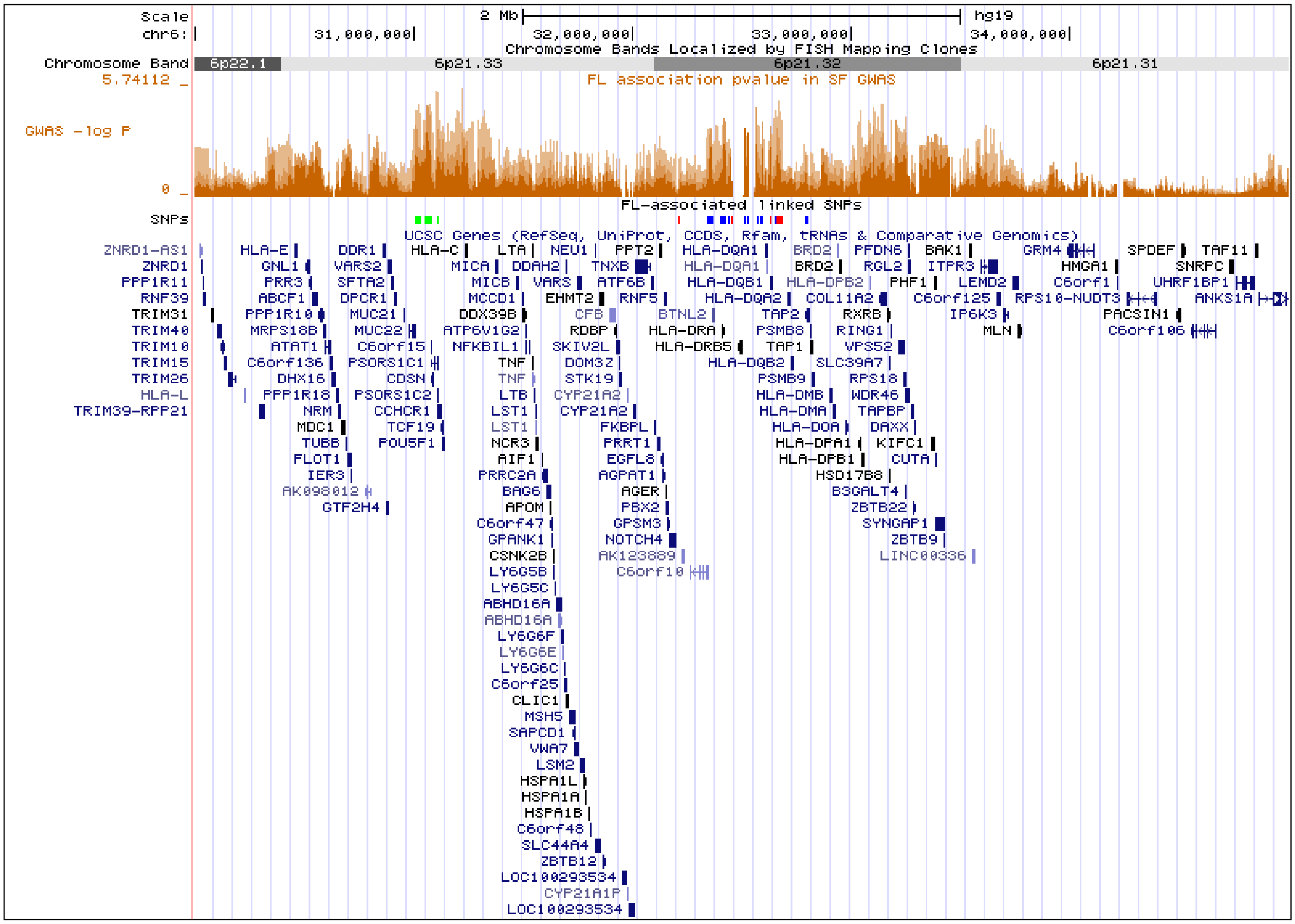

**Figure S5. Follicular Lymphoma (FL) Associated Variants in the 6p21.32-33 HLA Class II Region**

In the top track ('GWAS –log P'), association trend p-values (as –log10) obtained from a genome-wide association study of FL, are shown for SNPs in the region. In the 'SNPs' track, established FL-associated variants are shown, with rs6457327-linked SNPs colored in green, rs10484561-linked SNPs in blue and rs2647012-linked SNPs in red.

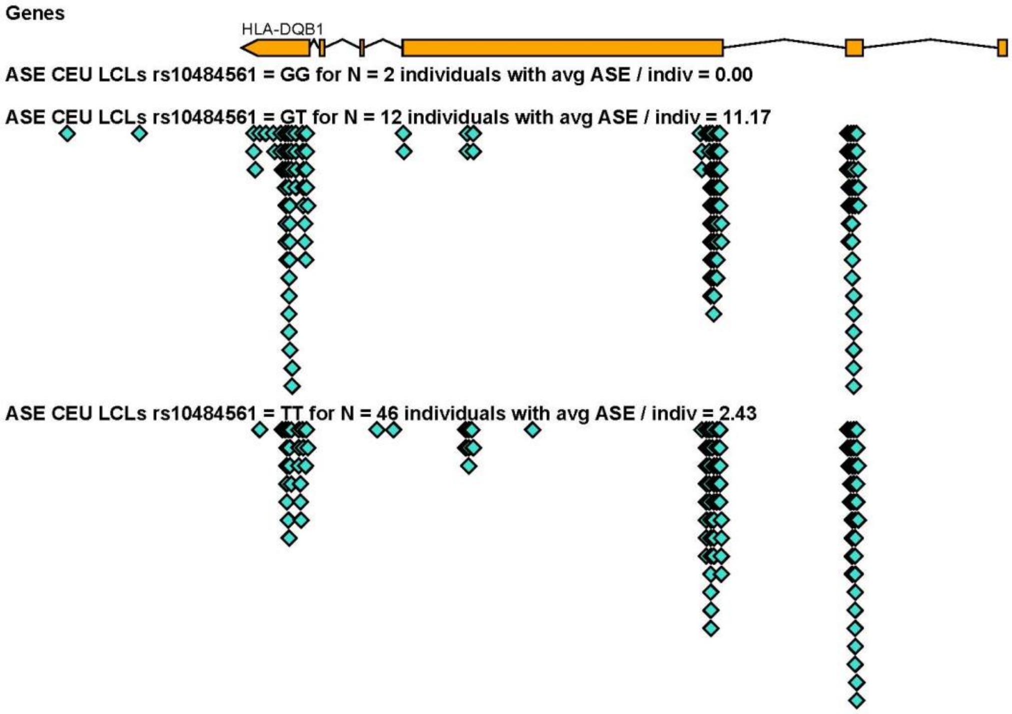

**Figure S6. Representation of the Total Number of Significant *HLA-DQB1* ASE Sites Stratified by rs10484561 Genotype**

**Table S1**. **Results from the eQTL Analysis of the Three Follicular Lymphoma Associated Variants rs2647012, rs10484561, rs6457327, and Variants in LD on Two RNA-Seq Data Sets**

| Locus | Probe | | SNP | Loc (GRCh37) | $r^2$ | a/A | FL GWAS | | | RNA-Seq expression Spearman's BH p-value | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | Gene | Transcript | | | | | MAFca/co | Genotyped / Imputed | Trend p-value | GSE16921 | E-MTAB-197 |
| rs2647012 | HLA-DQA1 | ENST00000343139 | rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | - | 4.19e-03(h) |
| | | | rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | - | 5.83e-03(h) |
| | | | rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | - | 7.37e-03(h) |
| | | | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | - | 3.99e-03(h) |
| | | | rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | - | 4.66e-03(h) |
| | | | rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | - | 7.31e-03(h) |
| | | | rs2858332 | 6:32681161 | 0.72 | G/T | 0.39/0.47 | Imp | 3.90E-03 | - | 4.19e-03(h) |
| | | | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | - | 1.22e-05(h) |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | - | 1.22e-05(h) |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | - | 4.96e-03(h) |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | - | 4.96e-03(h) |
| | | ENST00000374949 | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | - | 2.14e-03(h) |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | - | 2.14e-03(h) |
| | | ENST00000395364 | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | - | 3.94e-06(h) |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | - | 3.94e-06(h) |
| | | NM_002122 | rs1612904 | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | 2.53e-03(h) | - |
| | | | rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | 8.45e-04(h) | - |
| | | | rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | 8.45e-04(h) | - |
| | | | rs2647040 | 6:32667280 | 1 | A/G | 0.29/0.36 | Imp | 6.11E-03 | 3.81e-04(h) | - |
| | | | rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | 8.45e-04(h) | - |
| | | | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | 8.45e-04(h) | - |
| | | | rs2856725 | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | 1.48e-03(h) | - |
| | | | rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | 8.45e-04(h) | - |
| | | | rs2858310 | 6:32668323 | 1 | G/A | 0.28/0.36 | Imp | 3.42E-03 | 3.81e-04(h) | - |
| | | | rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | 8.45e-04(h) | - |
| | | | rs3129719 | 6:32661779 | 0.67 | T/G | 0.13/0.21 | Imp | 2.92E-04 | 6.72e-05(h) | - |
| | | | rs3129720 | 6:32663631 | 0.69 | T/C | 0.13/0.21 | Imp | 3.55E-04 | 7.71e-05(h) | - |
| | | | rs3134975 | 6:32652581 | 0.67 | T/C | 0.14/0.23 | Imp | 2.62E-04 | 6.72e-05(h) | - |
| | | | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | 2.61e-05(h) | - |
| | | | rs3998157 | 6:32678477 | 0.89 | C/A | 0.30/0.39 | Imp | 7.16E-04 | 8.94e-03(h) | - |
| | | | rs4273729 | 6:32678597 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(h) | - |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | rs4947344 | 6:32677846 | 0.58 | T/C | 0.18/0.28 | Imp | 3.61E-05 | 1.16e-04(h) | - |
| | | | rs6932517 | 6:32678182 | 0.93 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 2.51e-03(h) | - |
| | | | rs9267955 | 6:32213150 | 0.55 | A/G | 0.09/0.13 | Imp | 1.08E-02 | 4.09e-06(h) | - |
| | | | rs9273440 | 6:32627561 | 0.62 | T/C | 0.14/0.22 | Imp | 1.16E-03 | 2.61e-05(h) | - |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | 2.61e-05(h) | - |
| | | | rs9275295 | 6:32663391 | 0.73 | G/A | 0.40/0.49 | Imp | 1.59E-03 | 7.66e-03(l) | - |
| | | | rs9275517 | 6:32674649 | 0.9 | A/G | 0.30/0.39 | Genot | 9.31E-04 | 3.11e-03(h) | - |
| | | | rs9275521 | 6:32674952 | 1 | T/C | 0.29/0.36 | Imp | 4.98E-03 | 3.81e-04(h) | - |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | 2.58e-03(h) | - |
| | | | rs9275565 | 6:32677938 | 0.9 | T/C | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(h) | - |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | 2.58e-03(h) | - |
| | | | rs9275573 | 6:32679146 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(h) | - |
| | | | rs9380318 | 6:32677669 | 0.9 | C/T | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(h) | - |
| | HLA-DQA2 | NM_020056 (ENST00000374940) | rs1612904 | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | 8.57e-04(l) | S0.05 |
| | | | **rs1964995** | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | **9.57e-04(l)** | **6.70e-03(l)** |
| | | | **rs2647003** | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | **4.20e-04(l)** | **6.71e-03(l)** |
| | | | **rs2647012** | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | **4.20e-04(l)** | **4.56e-03(l)** |
| | | | rs2647040 | 6:32667280 | 1 | A/G | 0.29/0.36 | Imp | 6.11E-03 | 5.76e-04(l) | - |
| | | | **rs2647046** | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | **4.20e-04(l)** | **9.47e-03(l)** |
| | | | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | 4.20e-04(l) | S0.05 |
| | | | **rs2856725** | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | **2.03e-03(l)** | **4.97e-03(l)** |
| | | | **rs2858305** | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | **4.20e-04(l)** | **6.11e-03(l)** |
| | | | rs2858310 | 6:32668323 | 1 | G/A | 0.28/0.36 | Imp | 3.42E-03 | 5.76e-04(l) | - |
| | | | **rs2858324** | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | **4.20e-04(l)** | **8.10e-03(l)** |
| | | | rs3998157 | 6:32678477 | 0.89 | C/A | 0.30/0.39 | Imp | 7.16E-04 | 3.11e-03(l) | - |
| | | | rs4273729 | 6:32678597 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(l) | - |
| | | | rs6932517 | 6:32678182 | 0.93 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 2.09e-04(l) | - |
| | | | rs9275295 | 6:32663391 | 0.73 | G/A | 0.40/0.49 | Imp | 1.59E-03 | 2.58e-03(h) | - |
| | | | rs9275517 | 6:32674649 | 0.9 | A/G | 0.30/0.39 | Genot | 9.31E-04 | 6.18e-04(l) | - |
| | | | rs9275521 | 6:32674952 | 1 | T/C | 0.29/0.36 | Imp | 4.98E-03 | 5.76e-04(l) | - |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | 5.87e-04(l) | S0.05 |
| | | | rs9275565 | 6:32677938 | 0.9 | T/C | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(l) | - |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | 5.87e-04(l) | S0.05 |
| | | | rs9275573 | 6:32679146 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(l) | - |
| | | | rs9380318 | 6:32677669 | 0.9 | C/T | 0.30/0.39 | Imp | 7.16E-04 | 9.73e-04(l) | - |
| | HLA-DQB1 | ENST00000399065 | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | - | 8.47e-08(h) |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | - | 8.47e-08(h) |

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | rs9469220 | 6:32658310 | 0.5 | A/G | 0.38/0.47 | Imp | 5.06E-04 | - | 7.45e-03(l) |
| | | ENST00000399074 | rs1612904 | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | - | 8.64e-03(h) |
| | | | rs1964995 | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | - | 7.45e-03(h) |
| | | | rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | - | 1.77e-03(h) |
| | | | rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | - | 1.60e-03(h) |
| | | | rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | - | 1.75e-03(h) |
| | | | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | - | 2.58e-03(h) |
| | | | rs2856725 | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | - | 2.14e-03(h) |
| | | | rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | - | 2.14e-03(h) |
| | | | rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | - | 1.77e-03(h) |
| | | | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | - | 1.90e-07(h) |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | - | 1.90e-07(h) |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | - | 2.98e-03(h) |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | - | 2.98e-03(h) |
| | | ENST00000399082 | rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | - | 6.96e-03(h) |
| | | | rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | - | 5.27e-03(h) |
| | | | rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | - | 2.58e-03(h) |
| | | | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | - | 6.96e-03(h) |
| | | | rs2856725 | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | - | 3.99e-03(h) |
| | | | rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | - | 3.99e-03(h) |
| | | | rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | - | 9.14e-03(h) |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | - | 2.14e-03(h) |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | - | 2.14e-03(h) |
| | | NM_002123 (ENST00000399084) | **rs1612904** | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | **3.81e-04(h)** | **2.66e-03(h)** |
| | | | rs1964995 | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | S0.05 | 3.14e-03(h) |
| | | | **rs2647003** | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | **1.62e-04(h)** | **1.40e-03(h)** |
| | | | **rs2647012** | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | **1.62e-04(h)** | **1.21e-03(h)** |
| | | | rs2647040 | 6:32667280 | 1 | A/G | 0.29/0.36 | Imp | 6.11E-03 | 7.44e-05(h) | - |
| | | | **rs2647046** | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | **1.62e-04(h)** | **2.11e-03(h)** |
| | | | **rs2856717** | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | **1.62e-04(h)** | **2.14e-03(h)** |
| | | | **rs2856725** | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | **5.11e-04(h)** | **2.14e-03(h)** |
| | | | **rs2858305** | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | **1.62e-04(h)** | **1.75e-03(h)** |
| | | | rs2858310 | 6:32668323 | 1 | G/A | 0.28/0.36 | Imp | 3.42E-03 | 7.44e-05(h) | - |
| | | | **rs2858324** | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | **1.62e-04(h)** | **1.77e-03(h)** |
| | | | rs2858332 | 6:32681161 | 0.72 | G/T | 0.39/0.47 | Imp | 3.90E-03 | 1.92e-03(h) | S0.05 |
| | | | rs3129719 | 6:32661779 | 0.67 | T/G | 0.13/0.21 | Imp | 2.92E-04 | 3.00e-11(h) | - |
| | | | rs3129720 | 6:32663631 | 0.69 | T/C | 0.13/0.21 | Imp | 3.55E-04 | 4.74e-11(h) | - |

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | rs3134975 | 6:32652581 | 0.67 | T/C | 0.14/0.23 | Imp | 2.62E-04 | 3.00e-11(h) | - |
| | | | **rs3135006** | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | **1.18e-10(h)** | **7.28e-08(h)** |
| | | | rs3998157 | 6:32678477 | 0.89 | C/A | 0.30/0.39 | Imp | 7.16E-04 | 3.64e-03(h) | - |
| | | | rs4273729 | 6:32678597 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | - |
| | | | rs4947344 | 6:32677846 | 0.58 | T/C | 0.18/0.28 | Imp | 3.61E-05 | 1.69e-08(h) | - |
| | | | rs6932517 | 6:32678182 | 0.93 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 4.98e-04(h) | - |
| | | | rs9267955 | 6:32213150 | 0.55 | A/G | 0.09/0.13 | Imp | 1.08E-02 | 5.52e-10(h) | - |
| | | | rs9273440 | 6:32627561 | 0.62 | T/C | 0.14/0.22 | Imp | 1.16E-03 | 1.18e-10(h) | - |
| | | | **rs9273448** | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | **1.18e-10(h)** | **7.28e-08(h)** |
| | | | rs9275517 | 6:32674649 | 0.9 | A/G | 0.30/0.39 | Genot | 9.31E-04 | 9.15e-04(h) | - |
| | | | rs9275521 | 6:32674952 | 1 | T/C | 0.29/0.36 | Imp | 4.98E-03 | 7.44e-05(h) | - |
| | | | **rs9275524** | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | **4.20e-04(h)** | **1.01e-03(h)** |
| | | | rs9275565 | 6:32677938 | 0.9 | T/C | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | - |
| | | | **rs9275572** | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | **4.20e-04(h)** | **1.01e-03(h)** |
| | | | rs9275573 | 6:32679146 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | - |
| | | | rs9275588 | 6:32680423 | 0.72 | A/G | 0.39/0.47 | Imp | 3.90E-03 | 9.73e-04(l) | - |
| | | | rs9380318 | 6:32677669 | 0.9 | C/T | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | - |
| | HLA-DQB2 | NR_003937 | rs3129719 | 6:32661779 | 0.67 | T/G | 0.13/0.21 | Imp | 2.92E-04 | 8.36e-05(l) | - |
| | | | rs3129720 | 6:32663631 | 0.69 | T/C | 0.13/0.21 | Imp | 3.55E-04 | 1.46e-04(l) | - |
| | | | rs3134975 | 6:32652581 | 0.67 | T/C | 0.14/0.23 | Imp | 2.62E-04 | 8.36e-05(l) | - |
| | | | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | 7.44e-05(l) | - |
| | | | rs4947344 | 6:32677846 | 0.58 | T/C | 0.18/0.28 | Imp | 3.61E-05 | 3.36e-05(l) | - |
| | | | rs9267955 | 6:32213150 | 0.55 | A/G | 0.09/0.13 | Imp | 1.08E-02 | 1.19e-03(l) | - |
| | | | rs9273440 | 6:32627561 | 0.62 | T/C | 0.14/0.22 | Imp | 1.16E-03 | 7.44e-05(l) | - |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | 7.44e-05(l) | - |
| | HLA-DRA | ENST00000374982 | rs1612904 | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | - | 1.77e-03(h) |
| | | | rs1964995 | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | - | 2.14e-03(h) |
| | | | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | - | 2.14e-03(h) |
| | | | rs6457617 | 6:32663851 | 0.67 | C/T | 0.40/0.49 | Genot | 1.47E-03 | - | 6.19e-03(h) |
| | | | rs6457620 | 6:32663999 | 0.67 | G/C | 0.40/0.49 | Imp | 1.74E-03 | - | 6.19e-03(h) |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | - | 2.14e-03(h) |
| | | | rs9275224 | 6:32659878 | 0.67 | A/G | 0.40/0.48 | Imp | 1.65E-03 | - | 6.19e-03(h) |
| | | | rs9275245 | 6:32660943 | 0.67 | A/G | 0.39/0.48 | Genot | 1.36E-03 | - | 6.19e-03(h) |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | - | 2.14e-03(h) |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | - | 2.14e-03(h) |
| | | NM_019111 (ENST00000395388) | rs1964995 | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | NS | 2.14e-03(l) |
| | | | rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | NS | 4.19e-03(l) |

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | NS | 3.21e-03(l) |
| | | | rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | NS | 4.19e-03(l) |
| | | | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | NS | 4.11e-03(l) |
| | | | rs2856725 | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | NS | 8.68e-03(l) |
| | | | rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | NS | 2.97e-03(l) |
| | | | rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | NS | 5.29e-03(l) |
| | | | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | NS | 4.76e-03(l) |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | NS | 4.76e-03(l) |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | NS | 2.08e-03(l) |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | NS | 2.08e-03(l) |
| | HLA-DRB1 | NM_002124 (ENST00000360004) | **rs1612904** | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | **4.36e-03(h)** | **7.33e-03(h)** |
| | | | rs1964995 | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | 8.37e-04(h) | S0.05 |
| | | | rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | 1.61e-03(h) | NS |
| | | | rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | 1.61e-03(h) | NS |
| | | | rs2647040 | 6:32667280 | 1 | A/G | 0.29/0.36 | Imp | 6.11E-03 | 4.20e-04(h) | - |
| | | | rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | 1.61e-03(h) | NS |
| | | | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | 1.61e-03(h) | NS |
| | | | rs2856725 | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | 1.65e-03(h) | NS |
| | | | rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | 1.61e-03(h) | NS |
| | | | rs2858310 | 6:32668323 | 1 | G/A | 0.28/0.36 | Imp | 3.42E-03 | 4.20e-04(h) | - |
| | | | rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | 1.61e-03(h) | NS |
| | | | rs3129719 | 6:32661779 | 0.67 | T/G | 0.13/0.21 | Imp | 2.92E-04 | 1.06e-04(h) | - |
| | | | rs3129720 | 6:32663631 | 0.69 | T/C | 0.13/0.21 | Imp | 3.55E-04 | 1.56e-04(h) | - |
| | | | rs3134975 | 6:32652581 | 0.67 | T/C | 0.14/0.23 | Imp | 2.62E-04 | 1.06e-04(h) | - |
| | | | **rs3135006** | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | **3.81e-04(h)** | **3.91e-05(h)** |
| | | | rs3998157 | 6:32678477 | 0.89 | C/A | 0.30/0.39 | Imp | 7.16E-04 | 2.96e-04(h) | - |
| | | | rs4273729 | 6:32678597 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.45e-04(h) | - |
| | | | rs4947344 | 6:32677846 | 0.58 | T/C | 0.18/0.28 | Imp | 3.61E-05 | 1.16e-04(h) | - |
| | | | rs6932517 | 6:32678182 | 0.93 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 2.26e-04(h) | - |
| | | | rs9267955 | 6:32213150 | 0.55 | A/G | 0.09/0.13 | Imp | 1.08E-02 | 2.55e-07(h) | - |
| | | | rs9273440 | 6:32627561 | 0.62 | T/C | 0.14/0.22 | Imp | 1.16E-03 | 3.81e-04(h) | - |
| | | | **rs9273448** | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | **3.81e-04(h)** | **3.91e-05(h)** |
| | | | rs9275517 | 6:32674649 | 0.9 | A/G | 0.30/0.39 | Genot | 9.31E-04 | 6.32e-04(h) | - |
| | | | rs9275521 | 6:32674952 | 1 | T/C | 0.29/0.36 | Imp | 4.98E-03 | 4.20e-04(h) | - |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | 5.60e-04(h) | NS |
| | | | rs9275565 | 6:32677938 | 0.9 | T/C | 0.30/0.39 | Imp | 7.16E-04 | 1.45e-04(h) | - |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | 5.60e-04(h) | NS |

|  |  |  |  |  |  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|---|---|---|---|---|
|  |  |  | rs9275573 | 6:32679146 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.45e-04(h) | - |
|  |  |  | rs9380318 | 6:32677669 | 0.9 | C/T | 0.30/0.39 | Imp | 7.16E-04 | 1.45e-04(h) | - |
|  | HLA-DRB5 | NM_002125 (ENST00000374975) | rs1612904 | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | NS | 9.47e-03(h) |
|  |  |  | rs3129719 | 6:32661779 | 0.67 | T/G | 0.13/0.21 | Imp | 2.92E-04 | 1.45e-04(h) | - |
|  |  |  | rs3129720 | 6:32663631 | 0.69 | T/C | 0.13/0.21 | Imp | 3.55E-04 | 2.28e-04(h) | - |
|  |  |  | rs3134975 | 6:32652581 | 0.67 | T/C | 0.14/0.23 | Imp | 2.62E-04 | 1.45e-04(h) | - |
|  |  |  | **rs3135006** | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | **4.49e-04(h)** | **3.13e-05(h)** |
|  |  |  | rs4947344 | 6:32677846 | 0.58 | T/C | 0.18/0.28 | Imp | 3.61E-05 | 7.80e-04(h) | - |
|  |  |  | rs9267955 | 6:32213150 | 0.55 | A/G | 0.09/0.13 | Imp | 1.08E-02 | 4.69e-09(h) | - |
|  |  |  | rs9273440 | 6:32627561 | 0.62 | T/C | 0.14/0.22 | Imp | 1.16E-03 | 4.49e-04(h) | - |
|  |  |  | **rs9273448** | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | **4.49e-04(h)** | **3.13e-05(h)** |
|  | HLA-DRB6 | NR_001298 | rs1612904 | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | 1.38e-05(l) | - |
|  |  |  | rs1964995 | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | 3.00e-11(l) | - |
|  |  |  | rs2395522 | 6:32664722 | 0.75 | T/A | 0.40/0.48 | Imp | 1.91E-03 | 4.09e-04(h) | - |
|  |  |  | rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | 3.83e-06(l) | - |
|  |  |  | rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | 3.83e-06(l) | - |
|  |  |  | rs2647040 | 6:32667280 | 1 | A/G | 0.29/0.36 | Imp | 6.11E-03 | 1.18e-06(l) | - |
|  |  |  | rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | 3.83e-06(l) | - |
|  |  |  | rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | 3.83e-06(l) | - |
|  |  |  | rs2856725 | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | 7.57e-05(l) | - |
|  |  |  | rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | 3.83e-06(l) | - |
|  |  |  | rs2858310 | 6:32668323 | 1 | G/A | 0.28/0.36 | Imp | 3.42E-03 | 1.18e-06(l) | - |
|  |  |  | rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | 3.83e-06(l) | - |
|  |  |  | rs2858332 | 6:32681161 | 0.72 | G/T | 0.39/0.47 | Imp | 3.90E-03 | 7.44e-05(l) | - |
|  |  |  | rs3129719 | 6:32661779 | 0.67 | T/G | 0.13/0.21 | Imp | 2.92E-04 | 8.15e-04(l) | - |
|  |  |  | rs3129720 | 6:32663631 | 0.69 | T/C | 0.13/0.21 | Imp | 3.55E-04 | 1.62e-03(l) | - |
|  |  |  | rs3134975 | 6:32652581 | 0.67 | T/C | 0.14/0.23 | Imp | 2.62E-04 | 8.15e-04(l) | - |
|  |  |  | rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | 6.63e-04(l) | - |
|  |  |  | rs3998157 | 6:32678477 | 0.89 | C/A | 0.30/0.39 | Imp | 7.16E-04 | 6.95e-08(l) | - |
|  |  |  | rs4248168 | 6:32659743 | 0.76 | G/C | 0.40/0.48 | Imp | 1.65E-03 | 5.76e-04(l) | - |
|  |  |  | rs4273729 | 6:32678597 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.78e-09(l) | - |
|  |  |  | rs4713581 | 6:32660023 | 0.74 | C/T | 0.40/0.48 | Imp | 1.65E-03 | 2.66e-04(h) | - |
|  |  |  | rs4713582 | 6:32660051 | 0.78 | T/C | 0.40/0.48 | Imp | 1.65E-03 | 5.76e-04(l) | - |
|  |  |  | rs4713583 | 6:32660153 | 0.79 | T/G | 0.40/0.48 | Imp | 1.65E-03 | 3.04e-03(l) | - |
|  |  |  | rs4947344 | 6:32677846 | 0.58 | T/C | 0.18/0.28 | Imp | 3.61E-05 | 3.89e-05(l) | - |
|  |  |  | rs5002702 | 6:32659158 | 0.76 | G/A | 0.40/0.48 | Imp | 1.58E-03 | 5.76e-04(l) | - |
|  |  |  | rs6457617 | 6:32663851 | 0.67 | C/T | 0.40/0.49 | Genot | 1.47E-03 | 3.81e-04(l) | - |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | rs6457620 | 6:32663999 | 0.67 | G/C | 0.40/0.49 | Imp | 1.74E-03 | 3.81e-04(l) | - |
| | | | rs6932517 | 6:32678182 | 0.93 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 3.23e-08(l) | - |
| | | | rs9267955 | 6:32213150 | 0.55 | A/G | 0.09/0.13 | Imp | 1.08E-02 | 8.32e-04(l) | - |
| | | | rs9273440 | 6:32627561 | 0.62 | T/C | 0.14/0.22 | Imp | 1.16E-03 | 6.63e-04(l) | - |
| | | | rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | 6.63e-04(l) | - |
| | | | rs9275224 | 6:32659878 | 0.67 | A/G | 0.40/0.48 | Imp | 1.65E-03 | 3.81e-04(l) | - |
| | | | rs9275245 | 6:32660943 | 0.67 | A/G | 0.39/0.48 | Genot | 1.36E-03 | 3.81e-04(l) | - |
| | | | rs9275292 | 6:32663289 | 0.76 | C/A | 0.40/0.48 | Imp | 1.52E-03 | 5.76e-04(l) | - |
| | | | rs9275295 | 6:32663391 | 0.73 | G/A | 0.40/0.49 | Imp | 1.59E-03 | 1.69e-03(h) | - |
| | | | rs9275516 | 6:32674643 | 0.84 | A/G | 0.30/0.39 | Imp | 9.31E-04 | 2.24e-05(l) | - |
| | | | rs9275517 | 6:32674649 | 0.9 | A/G | 0.30/0.39 | Genot | 9.31E-04 | 1.17e-08(l) | - |
| | | | rs9275521 | 6:32674952 | 1 | T/C | 0.29/0.36 | Imp | 4.98E-03 | 1.18e-06(l) | - |
| | | | rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | 5.28e-09(l) | - |
| | | | rs9275565 | 6:32677938 | 0.9 | T/C | 0.30/0.39 | Imp | 7.16E-04 | 1.78e-09(l) | - |
| | | | rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | 5.28e-09(l) | - |
| | | | rs9275573 | 6:32679146 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.78e-09(l) | - |
| | | | rs9275588 | 6:32680423 | 0.72 | A/G | 0.39/0.47 | Imp | 3.90E-03 | 1.45e-04(h) | - |
| | | | rs9380318 | 6:32677669 | 0.9 | C/T | 0.30/0.39 | Imp | 7.16E-04 | 1.78e-09(l) | - |
| rs6457327 | C6orf26 | NM_001039651 (ENST00000395817) | rs1064191 | 6:31075375 | 0.63 | C/T | 0.37/0.48 | Genot | 8.95E-05 | NS | 2.14e-03(h) |
| | | | rs1265063 | 6:31077938 | 0.63 | G/A | 0.37/0.47 | Imp | 1.53E-04 | NS | 2.14e-03(h) |
| | | | rs2233966 | 6:31080859 | 0.62 | G/A | 0.35/0.47 | Imp | 4.74E-05 | NS | 2.14e-03(h) |
| | | | rs2517450 | 6:31061439 | 0.63 | T/C | 0.37/0.48 | Genot | 1.00E-04 | NS | 2.14e-03(h) |
| | | | rs2517452 | 6:31060162 | 0.63 | C/T | 0.37/0.48 | Genot | 1.00E-04 | NS | 2.14e-03(h) |
| | | | rs2517453 | 6:31060115 | 0.63 | T/C | 0.37/0.48 | Imp | 1.00E-04 | NS | 2.14e-03(h) |
| | | | rs2517455 | 6:31059900 | 0.63 | C/T | 0.37/0.48 | Imp | 1.00E-04 | NS | 2.14e-03(h) |
| | | | rs2517532 | 6:31018407 | 0.6 | A/G | 0.31/0.42 | Genot | 5.80E-05 | NS | 7.31e-03(l) |
| | | | rs2517534 | 6:31017334 | 0.6 | A/G | 0.31/0.42 | Imp | 5.80E-05 | NS | 7.31e-03(l) |
| | | | rs2535283 | 6:31068420 | 0.63 | G/A | 0.37/0.48 | Imp | 8.95E-05 | NS | 2.14e-03(h) |
| | | | rs2535293 | 6:31062363 | 0.63 | G/A | 0.37/0.48 | Imp | 1.00E-04 | NS | 2.14e-03(h) |
| | | | rs2535294 | 6:31062133 | 0.61 | G/A | 0.37/0.48 | Imp | 1.00E-04 | NS | 2.14e-03(h) |
| | | | rs2535303 | 6:31055423 | 0.63 | A/G | 0.37/0.48 | Genot | 1.00E-04 | NS | 2.14e-03(h) |
| | | | rs2535310 | 6:31053257 | 0.62 | C/A | 0.37/0.48 | Genot | 1.37E-04 | NS | 2.14e-03(h) |
| | | | rs3130553 | 6:31082285 | 0.52 | G/A | 0.58/0.47 | Imp | 3.55E-04 | NS | 9.31e-03(h) |
| | | | rs3130554 | 6:31082304 | 0.52 | G/T | 0.58/0.47 | Imp | 3.55E-04 | NS | 9.31e-03(h) |
| | | | rs7381897 | 6:31073903 | 0.63 | G/A | 0.37/0.48 | Imp | 8.95E-05 | NS | 2.14e-03(h) |
| | HCG22 | NR_003948 | rs1265053 | 6:31079889 | 0.52 | C/G | 0.35/0.47 | Imp | 4.23E-05 | 7.39e-03(h) | - |
| | | | rs2517448 | 6:31062667 | 1 | T/C | 0.25/0.36 | Genot | 1.70E-05 | 4.98e-04(h) | - |

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | rs2517524 | 6:31025713 | 0.64 | A/C | 0.55/0.47 | Imp | 1.04E-02 | 8.57e-04(l) | - |
| | | | rs2517532 | 6:31018407 | 0.6 | A/G | 0.31/0.42 | Genot | 5.80E-05 | 3.12e-04(h) | - |
| | | | rs2517534 | 6:31017334 | 0.6 | A/G | 0.31/0.42 | Imp | 5.80E-05 | 3.12e-04(h) | - |
| | | | rs2517538 | 6:31013541 | 0.84 | C/G | 0.24/0.35 | Imp | 3.17E-05 | 1.97e-05(h) | - |
| | | | rs2517545 | 6:31009302 | 0.81 | A/G | 0.26/0.36 | Imp | 2.37E-04 | 8.91e-05(h) | - |
| | | | rs2517548 | 6:31008818 | 0.81 | C/T | 0.26/0.36 | Genot | 2.37E-04 | 8.91e-05(h) | - |
| | | | rs2517550 | 6:31008368 | 0.81 | C/T | 0.26/0.36 | Imp | 1.45E-04 | 8.91e-05(h) | - |
| | | | rs2517552 | 6:31007590 | 0.84 | C/T | 0.24/0.36 | Genot | 4.10E-05 | 1.97e-05(h) | - |
| | | | rs2517556 | 6:31068895 | 0.58 | G/C | 0.37/0.48 | Imp | 8.95E-05 | 3.78e-03(h) | - |
| | | | rs2523862 | 6:31018842 | 0.59 | A/C | 0.31/0.42 | Imp | 5.80E-05 | 7.48e-04(h) | - |
| | | | rs2523870 | 6:31014116 | 0.82 | C/T | 0.24/0.35 | Genot | 3.17E-05 | 1.97e-05(h) | - |
| | | | rs2523871 | 6:31014009 | 0.83 | A/T | 0.24/0.35 | Imp | 3.17E-05 | 1.84e-05(h) | - |
| | | | rs2523872 | 6:31012730 | 0.84 | T/C | 0.24/0.36 | Genot | 4.10E-05 | 1.97e-05(h) | - |
| | | | rs2523873 | 6:31012263 | 0.82 | G/C | 0.25/0.36 | Imp | 1.01E-04 | 6.61e-05(h) | - |
| | | | rs2844665 | 6:31006855 | 0.81 | T/C | 0.26/0.36 | Genot | 2.54E-04 | 8.91e-05(h) | - |
| | | | rs2844669 | 6:31006092 | 0.81 | T/C | 0.26/0.36 | Imp | 2.45E-04 | 8.91e-05(h) | - |
| | | | rs3131927 | 6:31012996 | 0.84 | T/C | 0.24/0.36 | Imp | 2.83E-05 | 1.97e-05(h) | - |
| | | | rs3132562 | 6:31048992 | 1 | G/A | 0.30/0.41 | Imp | 8.98E-05 | 1.45e-04(h) | - |
| | | | rs6457327 | 6:31074030 | NA | A/C | 0.25/0.36 | Genot | 1.51E-05 | 4.98e-04(h) | - |
| | | | rs6905484 | 6:31071254 | 1 | T/C | 0.25/0.36 | Imp | 1.57E-05 | 5.14e-04(h) | - |
| | HLA-B | NM_005514 (ENST00000376228) | rs1064191 | 6:31075375 | 0.63 | C/T | 0.37/0.48 | Genot | 8.95E-05 | NS | 4.85e-03(l) |
| | | | rs1265052 | 6:31080471 | 0.55 | C/T | 0.33/0.45 | Imp | 3.56E-05 | NS | 3.86e-03(l) |
| | | | rs1265063 | 6:31077938 | 0.63 | G/A | 0.37/0.47 | Imp | 1.53E-04 | NS | 4.85e-03(l) |
| | | | rs2233966 | 6:31080859 | 0.62 | G/A | 0.35/0.47 | Imp | 4.74E-05 | NS | 3.94e-03(l) |
| | | | rs2517448 | 6:31062667 | 1 | T/C | 0.25/0.36 | Genot | 1.70E-05 | NS | 3.02e-03(h) |
| | | | rs2517450 | 6:31061439 | 0.63 | T/C | 0.37/0.48 | Genot | 1.00E-04 | NS | 4.85e-03(l) |
| | | | rs2517452 | 6:31060162 | 0.63 | C/T | 0.37/0.48 | Genot | 1.00E-04 | NS | 4.85e-03(l) |
| | | | rs2517453 | 6:31060115 | 0.63 | T/C | 0.37/0.48 | Imp | 1.00E-04 | NS | 4.85e-03(l) |
| | | | rs2517455 | 6:31059900 | 0.63 | C/T | 0.37/0.48 | Imp | 1.00E-04 | NS | 4.85e-03(l) |
| | | | rs2517532 | 6:31018407 | 0.6 | A/G | 0.31/0.42 | Genot | 5.80E-05 | NS | 2.02e-03(h) |
| | | | rs2517534 | 6:31017334 | 0.6 | A/G | 0.31/0.42 | Imp | 5.80E-05 | NS | 2.02e-03(h) |
| | | | rs2517538 | 6:31013541 | 0.84 | C/G | 0.24/0.35 | Imp | 3.17E-05 | NS | 5.73e-03(h) |
| | | | rs2517545 | 6:31009302 | 0.81 | A/G | 0.26/0.36 | Imp | 2.37E-04 | NS | 2.47e-03(h) |
| | | | rs2517548 | 6:31008818 | 0.81 | C/T | 0.26/0.36 | Genot | 2.37E-04 | NS | 2.47e-03(h) |
| | | | rs2517550 | 6:31008368 | 0.81 | C/T | 0.26/0.36 | Imp | 1.45E-04 | NS | 2.47e-03(h) |
| | | | rs2517552 | 6:31007590 | 0.84 | C/T | 0.24/0.36 | Genot | 4.10E-05 | NS | 2.01e-03(h) |
| | | | rs2523862 | 6:31018842 | 0.59 | A/C | 0.31/0.42 | Imp | 5.80E-05 | NS | 2.45e-03(h) |

|  |  |  |  |  |  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|---|---|---|---|---|
|  |  |  | rs2523870 | 6:31014116 | 0.82 | C/T | 0.24/0.35 | Genot | 3.17E-05 | NS | 2.01e-03(h) |
|  |  |  | rs2523872 | 6:31012730 | 0.84 | T/C | 0.24/0.36 | Genot | 4.10E-05 | NS | 2.01e-03(h) |
|  |  |  | rs2535283 | 6:31068420 | 0.63 | G/A | 0.37/0.48 | Imp | 8.95E-05 | NS | 4.85e-03(l) |
|  |  |  | rs2535293 | 6:31062363 | 0.63 | G/A | 0.37/0.48 | Imp | 1.00E-04 | NS | 4.85e-03(l) |
|  |  |  | rs2535294 | 6:31062133 | 0.61 | G/A | 0.37/0.48 | Imp | 1.00E-04 | NS | 4.85e-03(l) |
|  |  |  | rs2535303 | 6:31055423 | 0.63 | A/G | 0.37/0.48 | Genot | 1.00E-04 | NS | 4.85e-03(l) |
|  |  |  | rs2535310 | 6:31053257 | 0.62 | C/A | 0.37/0.48 | Genot | 1.37E-04 | NS | 4.85e-03(l) |
|  |  |  | rs2844665 | 6:31006855 | 0.81 | T/C | 0.26/0.36 | Genot | 2.54E-04 | NS | 2.47e-03(h) |
|  |  |  | rs2844669 | 6:31006092 | 0.81 | T/C | 0.26/0.36 | Imp | 2.45E-04 | NS | 2.47e-03(h) |
|  |  |  | rs3130553 | 6:31082285 | 0.52 | G/A | 0.58/0.47 | Imp | 3.55E-04 | NS | 8.68e-03(l) |
|  |  |  | rs3130554 | 6:31082304 | 0.52 | G/T | 0.58/0.47 | Imp | 3.55E-04 | NS | 8.68e-03(l) |
|  |  |  | rs3131927 | 6:31012996 | 0.84 | T/C | 0.24/0.36 | Imp | 2.83E-05 | NS | 3.56e-03(h) |
|  |  |  | rs6457327 | 6:31074030 | NA | A/C | 0.25/0.36 | Genot | 1.51E-05 | NS | 3.02e-03(h) |
|  |  |  | rs7381897 | 6:31073903 | 0.63 | G/A | 0.37/0.48 | Imp | 8.95E-05 | NS | 4.85e-03(l) |

Only eQTL associations that exhibited a Benjamini-Hochberg corrected p-value <= 0.01 in at least one dataset are shown.
a/A = minor/Major allele, NS = non-significant (BH pvalue > 0.05), S0.05 = significant at a 0.01 < BH p-value < 0.05, (h) = higher expression, (l) = lower expression.

**Table S2**. **rs2647012-Linked SNPs Associated with Differential *HLA-DQB1* Expression, Sorted by FL-Association Trend p-Value**

| SNP | Pos (hg19) | r2 rs2647012 | a/A | FL GWAS | | | RNA-Seq expression Spearman's BH p-value* | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | MAFca/co | G/I | Trend p-value | HLA-DQB1 | HLA-DRB1 | HLA-DRB6 | HLA-DQA1 | HLA-DQA2 |
| rs4947344 | 6:32677846 | 0.58 | T/C | 0.18/0.28 | Imp | 3.61E-05 | 1.69e-08(h) | 1.16e-04(h) | 3.89e-05(l) | 1.16e-04(h) | S0.05 |
| rs3134975 | 6:32652581 | 0.67 | T/C | 0.14/0.23 | Imp | 2.62E-04 | 3.00e-11(h) | 1.06e-04(h) | 8.15e-04(l) | 6.72e-05(h) | S0.05 |
| rs3129719 | 6:32661779 | 0.67 | T/G | 0.13/0.21 | Imp | 2.92E-04 | 3.00e-11(h) | 1.06e-04(h) | 8.15e-04(l) | 6.72e-05(h) | S0.05 |
| rs3129720 | 6:32663631 | 0.69 | T/C | 0.13/0.21 | Imp | 3.55E-04 | 4.74e-11(h) | 1.56e-04(h) | 1.62e-03(l) | 7.71e-05(h) | S0.05 |
| rs9469220 | 6:32658310 | 0.5 | A/G | 0.38/0.47 | Imp | 5.06E-04 | 7.45e-03(l) | NS | NS | NS | NS |
| rs3135006 | 6:32667119 | 0.62 | T/C | 0.13/0.21 | Imp | 6.31E-04 | 1.18e-10(h) | 3.91e-05(h) | 6.63e-04(l) | 3.94e-06(h) | S0.05 |
| rs4273729 | 6:32678597 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | 1.45e-04(h) | 1.78e-09(l) | 9.73e-04(h) | 9.73e-04(l) |
| rs9275565 | 6:32677938 | 0.9 | T/C | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | 1.45e-04(h) | 1.78e-09(l) | 9.73e-04(h) | 9.73e-04(l) |
| rs9275573 | 6:32679146 | 0.9 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | 1.45e-04(h) | 1.78e-09(l) | 9.73e-04(h) | 9.73e-04(l) |
| rs9380318 | 6:32677669 | 0.9 | C/T | 0.30/0.39 | Imp | 7.16E-04 | 1.99e-04(h) | 1.45e-04(h) | 1.78e-09(l) | 9.73e-04(h) | 9.73e-04(l) |
| rs3998157 | 6:32678477 | 0.89 | C/A | 0.30/0.39 | Imp | 7.16E-04 | 3.64e-03(h) | 2.96e-04(h) | 6.95e-08(l) | 8.94e-03(h) | 3.11e-03(l) |
| rs9275572 | 6:32678999 | 0.9 | A/G | 0.30/0.39 | Genot | 7.16E-04 | 4.20e-04(h) | 5.60e-04(h) | 5.28e-09(l) | 2.58e-03(h) | 5.87e-04(l) |
| rs6932517 | 6:32678182 | 0.93 | C/G | 0.30/0.39 | Imp | 7.16E-04 | 4.98e-04(h) | 2.26e-04(h) | 3.23e-08(l) | 2.51e-03(h) | 2.09e-04(l) |
| rs9275524 | 6:32675109 | 0.9 | T/C | 0.30/0.39 | Imp | 9.31E-04 | 4.20e-04(h) | 5.60e-04(h) | 5.28e-09(l) | 2.58e-03(h) | 5.87e-04(l) |
| rs9275517 | 6:32674649 | 0.9 | A/G | 0.30/0.39 | Genot | 9.31E-04 | 9.15e-04(h) | 6.32e-04(h) | 1.17e-08(l) | 3.11e-03(h) | 6.18e-04(l) |
| rs9273440 | 6:32627561 | 0.62 | T/C | 0.14/0.22 | Imp | 1.16E-03 | 1.18e-10(h) | 3.81e-04(h) | 6.63e-04(l) | 2.61e-05(h) | S0.05 |
| rs9273448 | 6:32627747 | 0.62 | A/G | 0.14/0.21 | Genot | 1.18E-03 | 1.18e-10(h) | 3.91e-05(h) | 6.63e-04(l) | 3.94e-06(h) | S0.05 |
| rs2647012 | 6:32664458 | NA | T/C | 0.28/0.36 | Genot | 2.97E-03 | 1.62e-04(h) | 1.61e-03(h) | 3.83e-06(l) | 8.45e-04(h) | 4.20e-04(l) |
| rs2858305 | 6:32670464 | 0.98 | G/T | 0.28/0.36 | Imp | 3.07E-03 | 1.62e-04(h) | 1.61e-03(h) | 3.83e-06(l) | 8.45e-04(h) | 4.20e-04(l) |
| rs2858324 | 6:32660375 | 1 | A/G | 0.29/0.36 | Imp | 3.27E-03 | 1.62e-04(h) | 1.61e-03(h) | 3.83e-06(l) | 8.45e-04(h) | 4.20e-04(l) |
| rs2647046 | 6:32668336 | 0.98 | A/C | 0.28/0.36 | Imp | 3.42E-03 | 1.62e-04(h) | 1.61e-03(h) | 3.83e-06(l) | 8.45e-04(h) | 4.20e-04(l) |
| rs2858310 | 6:32668323 | 1 | G/A | 0.28/0.36 | Imp | 3.42E-03 | 7.44e-05(h) | 4.20e-04(h) | 1.18e-06(l) | 3.81e-04(h) | 5.76e-04(l) |
| rs2856717 | 6:32670308 | 0.98 | A/G | 0.28/0.36 | Imp | 3.80E-03 | 1.62e-04(h) | 1.61e-03(h) | 3.83e-06(l) | 8.45e-04(h) | 4.20e-04(l) |
| rs2858332 | 6:32681161 | 0.72 | G/T | 0.39/0.47 | Imp | 3.90E-03 | 1.92e-03(h) | S0.05 | 7.44e-05(l) | 4.19e-03(h) | S0.05 |
| rs9275588 | 6:32680423 | 0.72 | A/G | 0.39/0.47 | Imp | 3.90E-03 | 9.73e-04(l) | S0.05 | 1.45e-04(h) | NS | S0.05 |
| rs2856725 | 6:32666738 | 0.97 | C/T | 0.29/0.36 | Imp | 4.92E-03 | 5.11e-04(h) | 1.65e-03(h) | 7.57e-05(l) | 1.48e-03(h) | 2.03e-03(l) |
| rs9275521 | 6:32674952 | 1 | T/C | 0.29/0.36 | Imp | 4.98E-03 | 7.44e-05(h) | 4.20e-04(h) | 1.18e-06(l) | 3.81e-04(h) | 5.76e-04(l) |
| rs2647003 | 6:32664880 | 1 | T/G | 0.29/0.36 | Imp | 5.44E-03 | 1.62e-04(h) | 1.61e-03(h) | 3.83e-06(l) | 8.45e-04(h) | 4.20e-04(l) |
| rs2647040 | 6:32667280 | 1 | A/G | 0.29/0.36 | Imp | 6.11E-03 | 7.44e-05(h) | 4.20e-04(h) | 1.18e-06(l) | 3.81e-04(h) | 5.76e-04(l) |
| rs1964995 | 6:32449411 | 0.68 | T/C | 0.53/0.45 | Genot | 6.19E-03 | 3.14e-03(h) | 8.37e-04(h) | 3.00e-11(l) | S0.05 | 9.57e-04(l) |
| rs1612904 | 6:32669018 | 0.85 | C/A | 0.24/0.31 | Imp | 7.05E-03 | 3.81e-04(h) | 4.36e-03(h) | 1.38e-05(l) | 2.53e-03(h) | 8.57e-04(l) |
| rs9267955 | 6:32213150 | 0.55 | A/G | 0.09/0.13 | Imp | 1.08E-02 | 5.52e-10(h) | 2.55e-07(h) | 8.32e-04(l) | 4.09e-06(h) | S0.05 |

* = Lower BH p-value from the GSE16921 and E-MTAB-197 datasets, (h) = higher expression, (l) = lower expression, NS= non-significant (BH p-value > 0.05), S0.05 = significant at a 0.01 < BH p-value < 0.05

**Table S3.** ***HLA-DQB1*** **Allele-Specific Expression (ASE) Analysis for rs2647012-Linked SNPs, Sorted by ASE p-Value**

| SNP | Loc(hg19) | r2 | Genes(s) | FL GWAS trend_p-value | HLA-DQB1 (NM_002123) eQTL Spearman's BH p-value | | ASE p-value | ENCODE/UCSC[source] |
|---|---|---|---|---|---|---|---|---|
| | | | | | GSE16921 | E-MTAB-197 | | |
| rs9275292 | 6:32663289 | 0.76 | - | 1.52E-03 | 1.08E-02 | - | 9.58E-05 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs4947344 | 6:32677846 | 0.58 | *MTCO3P1* | 3.61E-05 | 1.69E-08 | - | 1.28E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs9275573 | 6:32679146 | 0.9 | *MTCO3P1* | 7.16E-04 | 1.99E-04 | - | 3.34E-04 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs9275565 | 6:32677938 | 0.9 | *MTCO3P1* | 7.16E-04 | 1.99E-04 | - | 3.34E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs9380318 | 6:32677669 | 0.9 | *MTCO3P1* | 7.16E-04 | 1.99E-04 | - | 3.34E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs2647003 | 6:32664880 | 1 | - | 5.44E-03 | 1.62E-04 | 1.40E-03 | 9.26E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- CEBPB binding site [3] |
| rs2856725 | 6:32666738 | 0.97 | - | 4.92E-03 | 5.11E-04 | 2.14E-03 | 9.26E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs2647012 | 6:32664458 | NA | - | 2.97E-03 | 1.62E-04 | 1.21E-03 | 9.26E-04 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: high significance) (Gm12891) [4]<br>- Open chromatin region - FAIRE (Gm12891) [7]<br>Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - H3k27me3 signal enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [5]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Other<br>- Association traits: Follicular lymphoma, Systemic lupus erythematosus [6] |
| rs2858324 | 6:32660375 | 1 | - | 3.27E-03 | 1.62E-04 | 1.77E-03 | 9.26E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k9me3 signal enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs9275521 | 6:32674952 | 1 | *MTCO3P1* | 4.98E-03 | 7.44E-05 | - | 9.65E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [5]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs2647040 | 6:32667280 | 1 | - | 6.11E-03 | 7.44E-05 | - | 9.65E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs2858305 | 6:32670464 | 0.98 | *MTCO3P1* | 3.07E-03 | 1.62E-04 | 1.75E-03 | 9.65E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs2858310 | 6:32668323 | 1 | - | 3.42E-03 | 7.44E-05 | - | 9.65E-04 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878, Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878, Gm12891, Gm12892) [11]<br>- DNaseI hypersensitive site (Gm06990) [12]<br>- DNaseI hypersensitive site (Gm06990, Gm12878) [18]<br>- DNase hypersensitive area [13]<br>Histone Modification:<br>- H2A.Z, H3k27me3, H3k04me3 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- H3k04me3 signal enrichment (Gm12865) [5]<br>- Chromatin state: Weak/poised enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Rad21, Yy1sc281 binding site (Gm12878) [8]<br>- CEBPB, CTCF binding sites [3]<br>- CTCF binding site (Gm12878, Gm12892) [9]<br>- Z143 Std, CTCF Std, Rad2 IgR, WHIP IgM, p300 Std, Mxi1 IgM, SMC3 IgM binding sites (Gm12878) [10] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| rs2856717 | 6:32670308 | 0.98 | *MTCO3P1* | 3.80E-03 | 1.62E-04 | 2.14E-03 | 9.65E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs2647046 | 6:32668336 | 0.98 | - | 3.42E-03 | 1.62E-04 | 2.11E-03 | 9.65E-04 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878, Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878, Gm12891, Gm12892) [11]<br>- DNaseI hypersensitive site (Gm06990) [12]<br>- DNaseI hypersensitive site (Gm06990, Gm12878) [18]<br>- DNase hypersensitive area [13]<br>Histone Modifications:<br>- H2A.Z, H3k27me, H3k04me3 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- H3k04me3 signal enrichment (Gm12865) [5]<br>- Chromatin state: Weak/poised enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Rad21, Yy1sc281 binding sites (Gm12878) [8]<br>- CEBPB, CTCF, RFX5, YY1 binding sites [3]<br>- CTCF binding site (Gm12878, Gm12892) [9]<br>- Z143 Std, CTCF Std, Rad2 IgR, WHIP IgM, p300 Std, Mxi1 IgM, SMC3 IgM, BHL4 IgM binding sites (Gm12878) [10] |
| rs2395522 | 6:32664722 | 0.75 | - | 1.91E-03 | 2.18E-02 | - | 9.91E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs9275224 | 6:32659878 | 0.67 | - | 1.65E-03 | 1.75E-02 | 7.84E-02 | 9.91E-04 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878) [4]<br>- Region of open chromatin (OC code= 3: low significance) (Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878) [11]<br>- DNase hypersensitive area [13]<br>Histone Modifications:<br>- H3k04me1, H3k27me3, H3k9me3 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Insulator (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Rad21, Yy1sc281, ATF2, FOXM1, PML, BCLAF1 binding sites (Gm12878) [8]<br>- POU2 binding site (Gm12891) [8]<br>- SMC3, Znf143, ELF1, YY1, Rad21, RFX5, CTCF, GATA3 |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | binding sites [3]<br>- CTCF binding site (Gm12878, Gm12892) [9]<br>- Z143 Std, CTCF Std, Rad2 IgR, EBF1 IgM, STA1 Std, SMC3 IgM binding sites (Gm12878) [10]<br>- CTCF binding site (Gm12801) [14] |
| rs6457617 | 6:32663851 | 0.67 | - | 1.47E-03 | 1.75E-02 | 7.84E-02 | 9.91E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Other:<br>- Association traits: Systemic sclerosis, Graves' disease, Rheumatoid arthritis [6] |
| rs9275245 | 6:32660943 | 0.67 | - | 1.36E-03 | 1.75E-02 | 7.84E-02 | 9.91E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k9me3, H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs4713581 | 6:32660023 | 0.74 | - | 1.65E-03 | 1.36E-02 | - | 9.91E-04 | Chromatin Accessibility:<br>- Region of open chromatin (OC code=1: validated) (Gm12878) [4]<br>- Region of open chromatin (OC code=3: low significance) (Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12878, Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878) [11]<br>- DNaseI hypersensitive site (Gm12865) [12]<br>- DNaseI hypersensitive site (Gm06990, Gm12878, Gm12865, Gm12864) [18]<br>- DNase hypersensitive area [13]<br>Histone Modifications:<br>- H3K9me3, H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Insulator (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Rad21, Yylsc281, ATF2, FOXM1, PML, BCLAF1 binding sites (Gm12878) [8]<br>- Yy1sc281, POU2 binding sites (Gm12891) [8]<br>- TR4, CTCF, SMC3, Znf143, ELF1, YY1, Rad21, RFX5, GATA3 binding sites [3]<br>- CTCF binding site (Gm12878, Gm12891, Gm12892) [9]<br>- RFX5 IgM, EBF1 IgM, WHIP IgM, CTCF Std, Z143 Std, STA1 Std, Rad2 IgR, SMC3 IgM binding sites (Gm12878) [10]<br>- CTCF binding site (Gm06990, Gm12801, Gm12864, Gm12865, Gm12872, Gm12873, Gm12874, Gm12875, Gm12878) [14] |
| rs4713582 | 6:32660051 | 0.78 | - | 1.65E-03 | 1.08E-02 | - | 9.91E-04 | Chromatin Accessibility: |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - Region of open chromatin (OC code=1: validated) (Gm12878) [4]<br>- Region of open chromatin (OC code=3: low significance) (Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12878, Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878) [11]<br>- DNaseI hypersensitive site (Gm12865) [12]<br>- DNaseI hypersensitive site (Gm06990, Gm12878, Gm12865) [18]<br>- DNase hypersensitive area [13]<br>Histone Modifications:<br>- H3K9me3, H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Insulator (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Rad21, Yylsc281, ATF2, FOXM1, PML, BCLAF1 binding sites (Gm12878) [8]<br>- Yy1sc281, POU2 binding sites (Gm12891) [8]<br>- TR4, CTCF, SMC3, Znf143, ELF1, YY1, Rad21, RFX5, GATA3 binding sites [3]<br>- CTCF binding site (Gm12878, Gm12891, Gm12892) [9]<br>- RFX5 IgM, EBF1 IgM, WHIP IgM, CTCF Std, Z143 Std, STA1 Std, Rad2 IgR, SMC3 IgM binding sites (Gm12878) [10]<br>- CTCF binding site (Gm06990, Gm12801, Gm12864, Gm12865, Gm12872, Gm12873, Gm12874, Gm12875, Gm12878) [14] |
| rs6457620 | 6:32663999 | 0.67 | - | 1.74E-03 | 1.75E-02 | 7.84E-02 | 9.91E-04 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Other:<br>- Association traits: Rheumatoid arthritis [6] |
| rs4713583 | 6:32660153 | 0.79 | - | 1.65E-03 | 4.34E-02 | - | 9.91E-04 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878) [4]<br>- Region of open chromatin (OC code= 3: low significance) (Gm12891) [4]<br>- Open chromatin region - FAIRE (Gm12878, Gm12891) [7]<br>Histone Modifications:<br>- H3k9me3, H3k27me3 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Insulator (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Rad21, ATF2, PML, BCLAF1 binding sites (Gm12878) [8]<br>- TR4, CTCF, RFX5 binding sites [3]<br>- CTCF binding site (Gm12878, Gm12892) [9] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - CTCF Std, Z143 Std, Rad2 IgR, SMC3 IgM binding sites (Gm12878) [10] |
| rs4248168 | 6:32659743 | 0.76 | - | 1.65E-03 | 1.08E-02 | - | 9.91E-04 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: high significance) (Gm12878) [4]<br>- Open chromatin region - FAIRE (Gm12878) [7]<br>- DNaseI hypersensitive site (Gm06990) [18]<br>Histone Modifications:<br>- H3k9me3, H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak/poised enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- ATF2, PML, BCLAF1 binding sites (Gm12878) [8]<br>- SMC3 IgM binding site (Gm12878) [10] |
| rs9275295 | 6:32663391 | 0.73 | - | 1.59E-03 | 1.23E-02 | - | 1.18E-03 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs5002702 | 6:32659158 | 0.76 | - | 1.58E-03 | 1.08E-02 | - | 1.33E-03 | Histone Modifications:<br>- H3k9me3, H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1] - Chromatin state: Weak/poised enhancer (Gm12878) [2] |
| Rs9273448 | 6:32627747 | 0.62 | *HLA-DQB1-AS1, HLA-DQB1, XXbac-BPG254F23.6* | 1.18E-03 | 1.18E-10 | 7.28E-08 | 1.33E-03 | Chromatin Accessibility:<br>- Region of open chromatin (OC code=1: validated) (Gm12891) [4]<br>- Region of open chromatin (OC code=3: high significance) (Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12891) [11]<br>- DNaseI hypersensitive site (Gm06990, Gm12865) [12]<br>- DNaseI hypersensitive site (Gm06990, Gm12878, Gm12865, Gm12864) [18]<br>- DNase hypersensitive area [13]<br>Histone Modifications:<br>- H3K36me3, H3k04me1, H3k79me2, H3k9ac, H3k27ac, H3k4me2, H3k36me3, H3k04me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Strong enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2, NFATC1, EBF1, TCF12, BCLAF1, RUNX3, MTA3, POU2, PML, EBF1, TAF1, PBX3, TCF3 binding sites (Gm12878) [8]<br>- POL2, POU2, TAF1 binding sites (Gm12891) [8]<br>- POL2 binding site (Gm12892) [8] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - TBP, ELF1 binding site [3]<br>- Pol2 binding site (Gm12878, Gm12891) [9]<br>- PolS IgM, TBP IgM, Mxi1 IgM, CHD2 IgM, BHL4 IgM, STA1 Std, EBF1 Std, WHIP IgM, CTCF Std binding sites (Gm12878) [10]<br>RNA Binding Proteins:<br>- PABPC1 RNA binding site (Gm12878) [15][16]<br>- RipInput RNA binding site (Gm12878) [15][16] |
| rs9273440 | 6:32627561 | 0.62 | *HLA-DQB1-AS1, HLA-DQB1, XXbac-BPG254F23.6* | 1.16E-03 | 1.18E-10 | - | 1.49E-03 | Histone Modifications:<br>- H3k4me2, H3k04me1, H3k36me3, H3k79me2, H3k9ac, H3k36me3, H3k27ac, signal enrichment (Gm12878) [1]<br>- H3k4me3 signal enrichment (Gm12865) [5]<br>- Chromatin state: Strong enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2, NFATC1, MTA3, POU2, BCLAF1, PML, RUNX3 binding sites (Gm12878) [8]<br>- POL2 binding site (Gm12891, Gm12892) [8]<br>- Pol2-4H8 binding site [3]<br>- Pol2 binding site (Gm12878) [9]<br>- PolS IgM, EBF1 Std binding sites (Gm12878) [10]<br>- Pol2 IgM binding site (Gm12891) [10]<br>RNA Binding Proteins:<br>- PABPC1 RNA binding site (Gm12878) [15]<br>- RipInput RNA binding site (Gm12878) [15] |
| rs9275517 | 6:32674649 | 0.9 | *MTCO3P1* | 9.31E-04 | 9.15E-04 | - | 2.10E-03 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2]<br>Other:<br>- Vertebrate conserved element [17] |
| rs9275524 | 6:32675109 | 0.9 | *MTCO3P1* | 9.31E-04 | 4.20E-04 | 1.01E-03 | 2.10E-03 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs6932517 | 6:32678182 | 0.93 | *MTCO3P1* | 7.16E-04 | 4.98E-04 | - | 2.10E-03 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs9275516 | 6:32674643 | 0.84 | *MTCO3P1* | 9.31E-04 | 1.06E-01 | - | 2.10E-03 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2]<br>Other:<br>- Vertebrate conserved element [17] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| rs3998157 | 6:32678477 | 0.89 | *MTCO3P1* | 7.16E-04 | 3.64E-03 | - | 2.10E-03 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs9275572 | 6:32678999 | 0.9 | *MTCO3P1* | 7.16E-04 | 4.20E-04 | 1.01E-03 | 2.10E-03 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2]<br>Other:<br>- Association traits: Hepatocellular carcinoma, Alopecia areata [6] |
| rs1964995 | 6:32449411 | 0.68 | - | 6.19E-03 | 1.33E-02 | 3.14E-03 | 2.36E-03 | Histone Modifications:<br>- H2A.Z, H3k04me1, H3k9me3, H3k27ac signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs4273729 | 6:32678597 | 0.9 | *MTCO3P1* | 7.16E-04 | 1.99E-04 | - | 3.33E-03 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs9275588 | 6:32680423 | 0.72 | - | 3.90E-03 | 9.73E-04 | - | 1.74E-02 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs2858332 | 6:32681161 | 0.72 | *XXbac-BPG254F23.7* | 3.90E-03 | 1.92E-03 | 1.03E-02 | 1.74E-02 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak/poised enhancer (Gm12878) [2] |
| rs9469220 | 6:32658310 | 0.5 | - | 5.06E-04 | 3.18E-01 | 1.26E-01 | 3.04E-02 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: low significance) (Gm12891) [4]<br>- Open chromatin region - FAIRE (Gm12891) [7]<br>Histone Modifications:<br>- H3k9ac, H3k27ac, H3k9me3, H2A.Z, H3k27me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak/poised enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- PML binding site (Gm12878) [8]<br>- RFX5 binding site [3]<br>- BHL4 IgM, NYA IgM, RFX5 IgM, TBP IgM, Z143 Std, IRF3 IgM, NYB IgM, CHD2 IgM binding sites (Gm12878) [10]<br>Other:<br>- Association traits: Crohn's disease [6] |
| rs9267955 | 6:32213150 | 0.55 | - | 1.08E-02 | 5.52E-10 | - | 5.67E-02 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878, Gm12891) [4]<br>- DNaseI Hypersensitive zone (Gm12891, Gm12891) [11]<br>Histone Modifications:<br>- H3k4me2, H3k04me1 signal enrichment (Gm12878) [1] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak/poised enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- MTA3 binding site (Gm12878) [8]<br>- Mxi1 IgM, BHL4 IgM, SMC3 IgM binding sites (Gm12878) [10] |
| rs3129720 | 6:32663631 | 0.69 | - | 3.55E-04 | 4.74E-11 | - | 2.79E-01 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs3129719 | 6:32661779 | 0.67 | - | 2.92E-04 | 3.00E-11 | - | 2.79E-01 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k9me3, H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs3135006 | 6:32667119 | 0.62 | - | 6.31E-04 | 1.18E-10 | 7.28E-08 | 2.79E-01 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs1612904 | 6:32669018 | 0.85 | *MTCO3P1* | 7.05E-03 | 3.81E-04 | 2.66E-03 | 2.87E-01 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs3134975 | 6:32652581 | 0.67 | - | 2.62E-04 | 3.00E-11 | - | 4.58E-01 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: high significance) (Gm12891) [4]<br>- Open chromatin region - FAIRE (Gm12891) [7]<br>Histone Modifications:<br>- H3k04me1, H3k04me3, H3k9me3, H3k27me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak/poised enhancer (Gm12878) [2] |

The potential functional effect of the eQTLs was assessed by exploring their functional context using data from ENCODE and other regulatory datasets downloaded from the UCSC Genome Browser. Analyzed data from ENCODE include information on lymphoblastoid cell lines on chromatin accessibility (Duke DNaseI HS, Open Chrom Synth, UNC FAIRE, UW DNaseI DGF, UW DNaseI HS, DNase Clusters), chromatin interactions (UMass 5C), DNA methylation (HAIB Methyl RRBS, HAIB Methyl450), histone modifications (Broad ChromHMM, Broad Histone, UW Histone), RNA binding proteins (SUNY RIP GeneST, SUNY RIP-seq, SUNY Tiling), transcription factor binding sites (HAIB TFBS, SYDH TFBS, UTA TFBS, UW CTCF Binding, Txn Factor ChIP), and replication timing (UW Repli-seq). Other regulatory datasets analyzed include VISTA enhancers, conserved TFBSs from Transfac, conserved microRNA regulatory target sites as predicted by TargetScanHuman 5.1, transcription start sites from SwitchDB, regulatory elements from ORegAnno, and CpG islands. Additionally, we searched for overlap of eQTLs with vertebrate conserved elements and with trait-associated SNPs listed in the GWAS catalog.

Source: [1] Broad Histone; [2] Broad chromHMM; [3] Txn Factor ChIP; [4] Open Chrom Synth; [5] UW Histone; [6] GWAS catalog; [7] UNC FAIRE; [8] HAIB TFBS; [9] UTA TFBS; [10] SYDH TFBS; [11] Duke DNaseI HS; [12] UW DNaseI DGF; [13] DNase Clusters; [14] UW CTCF Binding; [15] SUNYRIPGeneST; [16] SUNYRIPseq; [17] UCSC Browser; [18] UW DNaseI HS; [19] HAIB Methyl450; [20] UW Repli-seq

**Table S4**. ***HLA-DQB1* Allele-Specific Expression (ASE) Analysis for rs10484561-Linked SNPs, Sorted by ASE p-Value**

| SNP | Loc(hg19) | r2 | Gene(s) | FL GWAS Trend_p | HLA-DQB1 (NM_002123) eQTL Spearman's BH p-value | | ASE p-value | ENCODE/UCSC [source] |
|---|---|---|---|---|---|---|---|---|
| | | | | | GSE16921 | E-MTAB-197 | | |
| rs17496307 | 6:32401036 | 0.84 | - | 2.01E-03 | 9.93E-01 | - | 2.62E-02 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: high significance) (Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12892) [7]<br>Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H2A.Z, H3k9me3, H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak transcribed (Gm12878) [2] |
| rs17496549 | 6:32409708 | 0.83 | *HLA-DRA* | 2.02E-03 | 9.91E-01 | - | 2.62E-02 | Histone Modifications:<br>- H3k9me3, H3k27ac, H3k04me3, H3k04me1, H3k4me2, H3k79me2, H3k9ac signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- H3k4me3 signal enrichment (Gm12878) [5]<br>- Chromatin state: Active Promoter (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Pol2-4H8 binding site [3]<br>- POL2, MTA3, BCLAF1, ATF2, STAT5A, RUNX3, PML, NFATC1, FOXM1 binding sites (Gm12878) [8]<br>- POL2, PAX5 binding sites (Gm12891) [8]<br>- POL2 binding site (Gm12892) [8]<br>- PolS IgM binding site (Gm12878) [10]<br>- Pol2 IgM binding site (Gm12891, Gm12892) [10] |
| rs35571839 | 6:32512613 | 0.8 | - | 2.60E-04 | 9.91E-01 | - | 2.79E-02 | Histone Modifications:<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs28895095 | 6:32418848 | 0.52 | - | 1.68E-04 | 9.63E-01 | 9.35E-01 | 3.01E-02 | Histone Modifications:<br>- H3k9me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs3177928 | 6:32412435 | 0.52 | *HLA-DRA* | 1.93E-04 | 9.63E-01 | 9.35E-01 | 3.25E-02 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: high significance) (Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12892) [7]<br>Histone Modifications:<br>- H3k9me3, H3k04me3, - H3k04me1, H3k79me2, H3k4me2, H3k36me3 signal enrichment (Gm12878) [1] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- H3k36me3 signal enrichment (Gm06990, Gm12878) [5]<br>- Chromatin state: Weak Promoter (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- Pol2-4H8,Pol2 binding sites [3]<br>- POL2 binding site (Gm12878, Gm12891, Gm12892) [8]<br>- MTA3, PML, ATF2, NFATC1, BCLAF1, STAT5A binding sites (Gm12878) [8]<br>- Pol2 binding site (Gm12878) [9]<br>- Mxi1IgM, PolS IgM binding sites (Gm12878) [10]<br>- Pol2 IgM binding site (Gm12891, Gm12892) [10]<br>RNA Binding Proteins:<br>- PABPC1, RipInput RNA binding sites (Gm12878) [16]<br>Other:<br>- Association traits: Total Cholesterol, LDL cholesterol [6] |
| rs13218331 | 6:32411362 | 0.52 | *HLA-DRA* | 1.89E-04 | 9.55E-01 | - | 3.25E-02 | Histone Modifications:<br>- H3k9me3, H3k27ac, H3k04me3, H3k04me1, H3k79me2, H3k4me2, H3k9ac, H3k36me3, H4k20me1 signal enrichment (Gm12878) [1]<br>- CTCF, EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak Promoter (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2 binding site (Gm12878, Gm12891, Gm12892) [8]<br>- MTA3, PML, POU2, NFATC1, BCLAF1, STAT5A binding sites (Gm12878) [8]<br>- PolS IgM binding site (Gm12878) [10]<br>- Pol2 IgM binding site (Gm12891, Gm12892) [10] |
| rs4530903 | 6:32581889 | 0.8 | - | 2.11E-04 | 9.91E-01 | 7.40E-01 | 3.45E-02 | Histone Modifications:<br>- H3k9me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2]<br>RNA Binding Proteins:<br>- PABPC1 RNA binding site (Gm12878) [15]<br>- RipInput RNA binding site (Gm12878) [15] |
| rs35954087 | 6:32440049 | 0.89 | *HLA-DRB9* | 1.92E-03 | 9.91E-01 | 8.95E-01 | 3.45E-02 | Histone Modifications:<br>- H3k9me3, H3k04me1, H2A.Z, H3k04me3, H3k27ac signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Promoter (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2 binding site (Gm12878, Gm12891, Gm12892) [8] |
| rs28605404 | 6:32569687 | 0.8 | - | 1.65E-04 | 9.91E-01 | 8.52E-01 | 3.45E-02 | Histone Modifications:<br>- H3k9me3, H3k79me2, H3k27ac, H3k04me1, H2A.Z signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Enhancer (Gm12878) [2]<br>RNA Binding Proteins:<br>- PABPC1, RipInput RNA binding site (Gm12878) [15] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| rs9391858 | 6:32341398 | 0.54 | *C6orf10* | 1.31E-03 | 9.63E-01 | 9.31E-01 | 3.45E-02 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Polycomb-repressed (Gm12878) [2] |
| rs241442 | 6:32797168 | 0.64 | *TAP2, XXbac-BPG246D15.9* | 6.04E-03 | 9.89E-01 | - | 3.76E-02 | Histone Modifications:<br>- H4k20me1, H3k79me2, H3k4me2, H3k36me3, H3k04me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Transcriptional elongation (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2 binding site (Gm12878) [8]<br>RNA Binding Proteins:<br>- PABPC1, SLBP RNA binding site (Gm12878) [15] |
| rs2857104 | 6:32790167 | 0.66 | *TAP2, XXbac-BPG246D15.9* | 1.19E-02 | 9.91E-01 | - | 3.94E-02 | Histone Modifications:<br>- H3k36me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2]<br>RNA Binding Proteins:<br>- SLBP RNA binding site (Gm12878) [15] |
| rs28895187 | 6:32424380 | 0.52 | *HLA-DRB9* | 1.68E-04 | 9.63E-01 | 9.35E-01 | 4.00E-02 | Histone Modifications:<br>- H3k9me3, H2A.Z, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs13209234 | 6:32415975 | 0.52 | *HLA-DRA* | 1.93E-04 | 9.76E-01 | 9.95E-01 | 4.00E-02 | Chromatin Accessibility:<br>- DNaseI hypersensitive site (Gm06990) [12]<br>Histone Modifications:<br>- H3k9me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- RUNX3, MEF2A, BATF, NFIC binding sites (Gm12878) [8]<br>- MEF2A, EBF binding sites [3]<br>- EBF1 Std, p300 Std binding sites (Gm12878) [10] |
| rs13212453 | 6:32422430 | 0.52 | - | 1.68E-04 | 9.90E-01 | 8.64E-01 | 4.00E-02 | Histone Modifications:<br>- H3k9me3, H2A.Z, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs28895171 | 6:32423021 | 0.52 | *HLA-DRB9* | 1.68E-04 | 9.63E-01 | 9.92E-01 | 4.00E-02 | Histone Modifications:<br>- H3k9me3, H2A.Z, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs12529093 | 6:32415858 | 0.52 | *HLA-DRA* | 1.93E-04 | 9.89E-01 | - | 4.00E-02 | Histone Modifications:<br>- H3k9me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- RUNX3, MEF2A, BATF, NFIC binding sites (Gm12878) [8]<br>- MEF2A, EBF binding sites [3]<br>- EBF1 Std, p300 Std binding sites (Gm12878) [10] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| rs2894253 | 6:32345540 | 0.71 | *C6orf10* | 3.03E-04 | 9.91E-01 | 9.36E-01 | 4.24E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Polycomb-repressed (Gm12878) [2] |
| rs6930777 | 6:32351566 | 0.71 | *C6orf10* | 2.72E-04 | 9.91E-01 | - | 4.24E-02 | Histone Modifications:<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs12528797 | 6:32345086 | 0.71 | *C6orf10* | 3.03E-04 | 9.91E-01 | 9.36E-01 | 4.24E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Polycomb-repressed (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- EBF1 Std binding site (Gm12878) [10] |
| rs12528368 | 6:32653643 | 0.87 | - | 8.50E-05 | 9.89E-01 | - | 4.33E-02 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878, Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12878, Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878, Gm12891, Gm12892) [11]<br>- DNaseI hypersensitive site (Gm12865) [12]<br>- DNaseI hypersensitive site (Gm06990, Gm12878, Gm12865, Gm12864) [18]<br>- DNase hypersensitive area [13]<br>Histone Modifications:<br>- H3k27me3, H3k27ac, H3k04me1, H3k9me3, H3k4me2, H3k04me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak Enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- STAT5A, FOXM1, MTA3, P300, RUNX3, ETS1, NFIC, PML, NFATC1, BATF, ATF2, POU2, TCF12, SP1, BCL11A, TCF3, SRF, MEFC, MEF2A, BCLAF1 binding sites (Gm12878) [8]<br>- PAX5, PU.1 binding site (Gm12878, Gm12891) [8]<br>- PU.1, RFX5(N-494), TBP, EBF, USF2, POU2F2, EBF1(C-8), PAX5-C20, Oct-2, p300, MEF2A, NFKB, SP1 binding sites [3]<br>- Z143 Std, RFX5 IgM, EBF1 IgM, STA3 Std, USF2 IgM, TBP IgM, SMC3 IgM, p300 IgM, p300 Std, Mxi1 IgM, NYB IgM, WHIP IgM, NFE2 Std, NYA IgM, IRF3 IgM, CHD2 IgM, BHL4 IgM binding sites (Gm12878) [10]<br>- TNF NKB IgR binding site (Gm12891) [10] |
| rs13198610 | 6:32425672 | 0.52 | *HLA-DRB9* | 1.68E-04 | 9.63E-01 | 9.35E-01 | 4.45E-02 | Histone Modifications:<br>- H3k9me3, H2A.Z, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs17427445 | 6:32663764 | 0.92 | - | 8.50E-05 | 9.82E-01 | - | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs6457614 | 6:32651900 | 0.92 | - | 8.50E-05 | 9.69E-01 | - | 5.63E-02 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: high significance) (Gm12891) [4]<br>- Open chromatin region - FAIRE (Gm12891) [7]<br>Histone Modifications:<br>- H3k27me3, H3k04me1, H3k9me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs10947332 | 6:32677440 | 0.92 | *MTCO3P1* | 1.36E-04 | 9.82E-01 | - | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs35998847 | 6:32666997 | 0.95 | - | 8.50E-05 | 9.91E-01 | 7.67E-01 | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs17212469 | 6:32665153 | 0.92 | - | 8.50E-05 | 9.82E-01 | - | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs17427431 | 6:32663644 | 0.91 | - | 8.50E-05 | 9.77E-01 | - | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs17427564 | 6:32667067 | 0.92 | - | 8.50E-05 | 9.82E-01 | - | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs17499411 | 6:32665002 | 0.91 | - | 8.50E-05 | 9.95E-01 | - | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- CEBPB binding site [3] |
| rs10484561 | 6:32665420 | NA | - | 8.50E-05 | 9.91E-01 | 8.04E-01 | 5.63E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Other:<br>- Association traits: Follicular lymphoma [6] |
| rs28530648 | 6:32527079 | 0.91 | *HLA-* | 4.24E-04 | 9.91E-01 | 8.09E-01 | 5.83E-02 | Histone Modifications: |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | *DRB6* | | | | | - H3k79me2 signal enrichment (Gm12878) [1]<br>- H3k4me3 signal enrichment (Gm12878, Gm12864) [5]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2 binding site (Gm12891) [8] |
| rs28895103 | 6:32419464 | 0.52 | - | 1.07E-04 | 9.45E-01 | 9.59E-01 | 6.80E-02 | Histone Modifications:<br>- H3k9me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs13192471 | 6:32671103 | 0.75 | *MTCO3P1* | 2.94E-03 | 9.85E-01 | 7.88E-01 | 8.68E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br>Other:<br>- Association traits: Rheumatoid arthritis [6] |
| rs35120848 | 6:32670495 | 0.75 | *MTCO3P1* | 2.94E-03 | 9.85E-01 | 7.88E-01 | 8.68E-02 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs7755224 | 6:32652317 | 1 | - | 5.59E-05 | 9.91E-01 | 8.81E-01 | 2.80E-01 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 3: high significance) (Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12891, Gm12892) [7]<br>Histone Modifications:<br>- H3k27me3, H3k04me1, H3k9me3 signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs35399661 | 6:32590990 | 0.62 | *HLA-DQA1* | 1.97E-03 | 9.89E-01 | 7.58E-01 | 6.35E-01 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878, Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12878, Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878, Gm12891, Gm12892) [11]<br>- DNaseI hypersensitive site (Gm06990, Gm12865) [12]<br>- DNaseI hypersensitive site (Gm06990, Gm12878, Gm12865, Gm12864) [18]<br>- DNase hypersensitive area [13]<br>Histone Modifications:<br>- H3k9me3, H2A.Z s, H3k04me3, H3k9ac, H3k4me2, H3k27ac signal enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- Chromatin state: Strong Enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2, POU2, STAT5A, FOXM1, MTA3, RUNX3, ETS1, NFIC, PML, NFATC1, BATF, ATF2, SP1, MEFC, MEF2A, BCLAF1, BCL3, Rad21, PAX5, IRF4, PU.1, TAF1 binding sites (Gm12878) [8] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | | | | | | - POL2, POU2 binding site (Gm12891) [8]<br>- POL2 binding site (Gm12892) [8]<br>- MEF2C_(SC-13268), RFX5 (N-494), TBP, NFKB, Oct-2, SP1, MEF2A, POU2F2, BCL11A binding sites [3]<br>- PolS IgM, STA1 Std, p300 Std, Rad2 IgR, EBF1 IgM, BHL4 IgM, STA3 Std, Mxi1 IgM, RFX5 IgM, NYA IgM, NFE2 Std, SMC3 IgM, TBP IgM, Z143 Std, p300 IgM, NYB IgM, CHD2 IgM, IRF3 IgM binding sites (Gm12878) [10]<br>- TNF NKB IgR, Pol2 IgM binding sites (Gm12891, Gm12892) [10]<br>RNA Binding Proteins:<br>- PABPC1, RipInput RNA binding sites (Gm12878) [15]<br>Other:<br>- Vertebrate conserved element [17] |
| rs17533090 | 6:32590722 | 0.62 | - | 1.97E-03 | 9.89E-01 | 7.58E-01 | 6.35E-01 | Chromatin Accessibility:<br>- Region of open chromatin (OC code= 1: validated) (Gm12878, Gm12891, Gm12892) [4]<br>- Open chromatin region - FAIRE (Gm12878, Gm12891, Gm12892) [7]<br>- DNaseI Hypersensitive zone (Gm12878, Gm12892) [11]<br>Histone Modifications:<br>- H3k9me3, H3k79me2, H2A.Z, H3k04me3, H3k04me1, H3k9ac, H3k4me2, H3k27ac signal enrichment (Gm12878) [1]<br>- CTCF binding enrichment (Gm12878) [1]<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k4me3 signal enrichment (Gm06990) [5]<br>- Chromatin state: Strong Enhancer (Gm12878) [2]<br>Transcription Factor Binding Sites:<br>- POL2, POU2, STAT5A, FOXM1, MTA3, RUNX3, ETS1, NFIC, PML, NFATC1, BATF, ATF2, SP1, MEFC, MEF2A, BCLAF1, BCL3, Rad21, PAX5, IRF4, PU.1, TAF1, YY1 binding sites (Gm12878) [8]<br>- POL2, POU2 binding site (Gm12891) [8]<br>- POL2 binding site (Gm12892) [8]<br>- Pol2 binding site [3]<br>- Pol2 binding site (Gm12878) [9]<br>- PolS IgM, p300 Std, Rad2 IgR, EBF1 IgM, BHL4 IgM, STA3 Std, Mxi1 IgM, SMC3 IgM, TBP IgM, Z143 Std, p300 IgM,, CHD2 IgM, IRF3 IgM binding sites (Gm12878) [10]<br>- TNF NKB IgR, Pol2 IgM binding sites (Gm12891, Gm12892) [10]<br>RNA Binding Proteins:<br>- PABPC1, RipInput RNA binding sites (Gm12878) [15] |
| rs17495626 | 6:32359643 | 0.65 | *HCG23, BTNL2* | 8.21E-04 | 9.89E-01 | - | 7.29E-01 | Histone Modifications:<br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs1794275 | 6:32671248 | 0.51 | *MTCO3* | 1.12E-02 | 9.91E-01 | - | 7.79E-01 | Histone Modifications: |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | *P1* | | | | | - EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H2A.Z signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2]<br><u>Transcription Factor Binding Sites:</u><br>- SRF binding site [3]<br><u>Other:</u><br>- Association traits: IgA nephropathy [6] |
| rs17208650 | 6:32358113 | 0.65 | *HCG23, BTNL2* | 1.20E-03 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Enhancer (Gm12878) [2] |
| rs17423649 | 6:32357133 | 0.6 | *HCG23, BTNL2, C6orf10* | 1.20E-03 | 9.84E-01 | 6.80E-01 | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs17208657 | 6:32358144 | 0.65 | *HCG23, BTNL2* | 1.20E-03 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Enhancer (Gm12878) [2] |
| rs17423691 | 6:32358345 | 0.65 | *HCG23, BTNL2* | 1.09E-03 | 9.71E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Enhancer (Gm12878) [2] |
| rs17495612 | 6:32359431 | 0.57 | *HCG23, BTNL2* | 8.21E-04 | 9.84E-01 | 6.80E-01 | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs17495592 | 6:32358533 | 0.65 | *HCG23, BTNL2* | 7.57E-04 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs17423753 | 6:32360341 | 0.65 | *HCG23, BTNL2* | 8.21E-04 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs12525722 | 6:32358163 | 0.65 | *HCG23, BTNL2* | 1.20E-03 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Enhancer (Gm12878) [2] |
| rs17208629 | 6:32357397 | 0.65 | *HCG23, BTNL2* | 1.20E-03 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| rs12529049 | 6:32357715 | 0.6 | *HCG23, BTNL2* | 1.20E-03 | 9.84E-01 | 6.80E-01 | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs1555115 | 6:32354520 | 0.65 | *HCG23, C6orf10, BTNL2* | 8.21E-04 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Heterochromatin; low signal (Gm12878) [2] |
| rs17423698 | 6:32358368 | 0.65 | *HCG23, BTNL2* | 7.57E-04 | 9.89E-01 | - | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Enhancer (Gm12878) [2] |
| rs3817969 | 6:32361388 | 0.57 | *HCG23, BTNL2* | 8.21E-04 | 9.84E-01 | 6.80E-01 | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2]<br><u>Transcription Factor Binding Sites:</u><br>- c-Fos, GATA-2, KAP1, Pol2(b) binding sites [3] |
| rs17202259 | 6:32357489 | 0.6 | *HCG23, BTNL2* | 1.20E-03 | 9.84E-01 | 6.80E-01 | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3, H3k04me1 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |
| rs16870123 | 6:32359460 | 0.6 | *HCG23, BTNL2* | 8.21E-04 | 9.84E-01 | 6.80E-01 | 7.92E-01 | <u>Histone Modifications:</u><br>- EZH2 binding enrichment (Gm12878) [1]<br>- H3k27me3 signal enrichment (Gm12878) [1]<br>- Chromatin state: Weak Transcribed (Gm12878) [2] |

The potential functional effect of the eQTLs was assessed by exploring their functional context using data from ENCODE and other regulatory datasets downloaded from the UCSC Genome Browser. Analyzed data from ENCODE include information on lymphoblastoid cell lines on chromatin accessibility (Duke DNaseI HS, Open Chrom Synth, UNC FAIRE, UW DNaseI DGF, UW DNaseI HS, DNase Clusters), chromatin interactions (UMass 5C), DNA methylation (HAIB Methyl RRBS, HAIB Methyl450), histone modifications (Broad ChromHMM, Broad Histone, UW Histone), RNA binding proteins (SUNY RIP GeneST, SUNY RIP-seq, SUNY Tiling), transcription factor binding sites (HAIB TFBS, SYDH TFBS, UTA TFBS, UW CTCF Binding, Txn Factor ChIP), and replication timing (UW Repli-seq). Other regulatory datasets analyzed include VISTA enhancers, conserved TFBSs from Transfac, conserved microRNA regulatory target sites as predicted by TargetScanHuman 5.1, transcription start sites from SwitchDB, regulatory elements from ORegAnno, and CpG islands. Additionally, we searched for overlap of eQTLs with vertebrate conserved elements and with trait-associated SNPs listed in the GWAS catalog.

Source: [1] Broad Histone; [2] Broad chromHMM; [3] Txn Factor ChIP; [4] Open Chrom Synth; [5] UW Histone; [6] GWAS catalog; [7] UNC FAIRE; [8] HAIB TFBS; [9] UTA TFBS; [10] SYDH TFBS; [11] Duke DNaseI HS; [12] UW DNaseI DGF; [13] DNase Clusters; [14] UW CTCF Binding; [15] SUNYRIPGeneST; [16] SUNYRIPseq; [17] UCSC Browser; [18] UW DNaseI HS; [19] HAIB Methyl450; [20] UW Repli-seq