

Supplementary Information

Chapter 2 Supplementary Information

GWAS summary statistics contributions

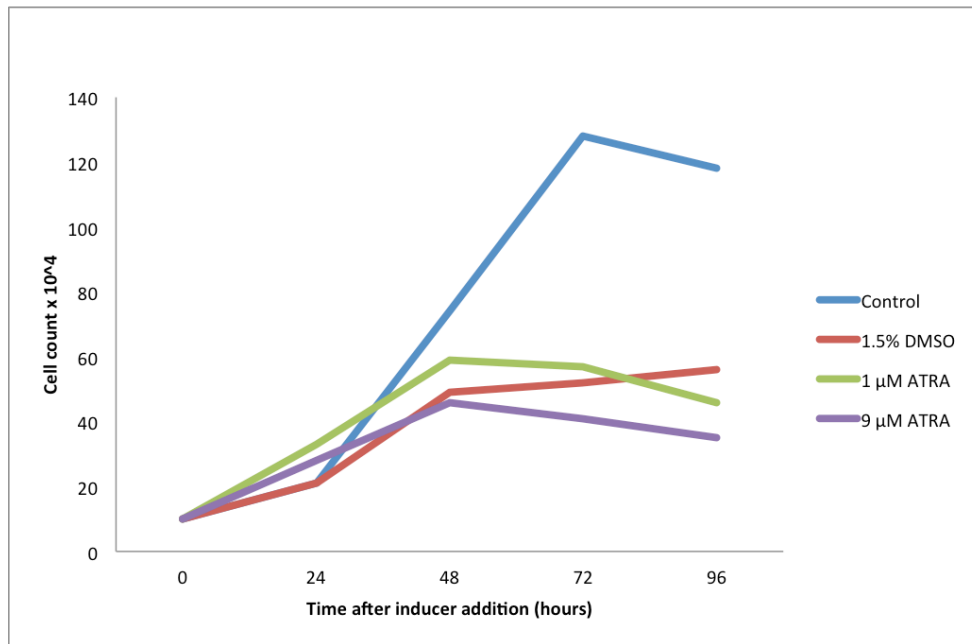
We thank the International Genomics of Alzheimer's Project (IGAP) for providing summary results data for these analyses. The investigators within IGAP contributed to the design and implementation of IGAP and/or provided data but did not participate in analysis or writing of this report. IGAP was made possible by the generous participation of the control subjects, the patients, and their families. The iSelect chips was funded by the French National Foundation on Alzheimer's disease and related disorders. EADI was supported by the LABEX (laboratory of excellence program investment for the future) DISTALZ grant, Inserm, Institut Pasteur de Lille, Universite de Lille 2 and the Lille University Hospital. GERAD was supported by the Medical Research Council (Grant no 503480), Alzheimer's Research UK (Grant no 503176), the Wellcome Trust (Grant no 082604/2/07/Z) and German Federal Ministry of Education and Research (BMBF): Competence Network Dementia (CND) grant no 01GI0102, 01GI0711, 01GI0420. CHARGE was partly supported by the NIH/NIA grant R01 AG033193 and the NIA AG081220 and AGES contract N01AG12100, the NHLBI grant R01 HL105756, the Icelandic Heart Association, and the Erasmus Medical Center and Erasmus University. ADGC was supported by the NIH/NIA grants: U01 AG032984, U24 AG021886, U01 AG016976, and the Alzheimer's Association grant ADGC10196728. International Genomics of Alzheimer's Project (IGAP) is a large two-stage study based upon genome-wide association studies (GWAS) on individuals of European ancestry. In stage 1, IGAP used genotyped and imputed data on 7,055,881 single nucleotide polymorphisms (SNPs) to meta-analyse four previously-published GWAS datasets consisting of 17,008 Alzheimer's disease cases and 37,154 controls (The European Alzheimer's disease Initiative EADI the Alzheimer Disease Genetics Consortium ADGC The Cohorts for Heart and Aging Research in Genomic Epidemiology consortium CHARGE The Genetic and Environmental Risk in AD consortium GERAD). In stage 2, 11,632 SNPs were genotyped and tested for association in an independent set of 8,572 Alzheimer's disease cases and 11,312 controls. Finally, a meta-analysis was performed combining results from stages 1 & 2. The text above was reproduced at request from the International Genomics of Alzheimer's Project (IGAP).

We thank the IAMDGC consortium for sharing the summary statistics and effect sizes of advanced age-related macular degeneration GWAS from Fritsche *et al.* (2016). The list of consortia members can be found in (Persad *et al.*, 2017).

Applications and queries can be made through the following contacts: goncalo@umich.edu, larsf@umich.edu

Further information on AMD statistics: <http://amdgenetics.org>

Chapter 2 Supplementary Figures



Supplementary Figure 2.1: Proliferation of dividing and differentiated HL60 cells Cell count of HL60 cells shown for each 24-hour period after addition of either DMSO or ATRA at different concentrations shown. The control HL60 cells had no addition and continued to proliferate until reaching a maximum, likely due to limited nutrients as medium was not changed in this period. The reduced proliferation of ATRA and DMSO conditions is evidence of differentiation.

Effects of ATRA & DMSO exposure on HL60 gene expression

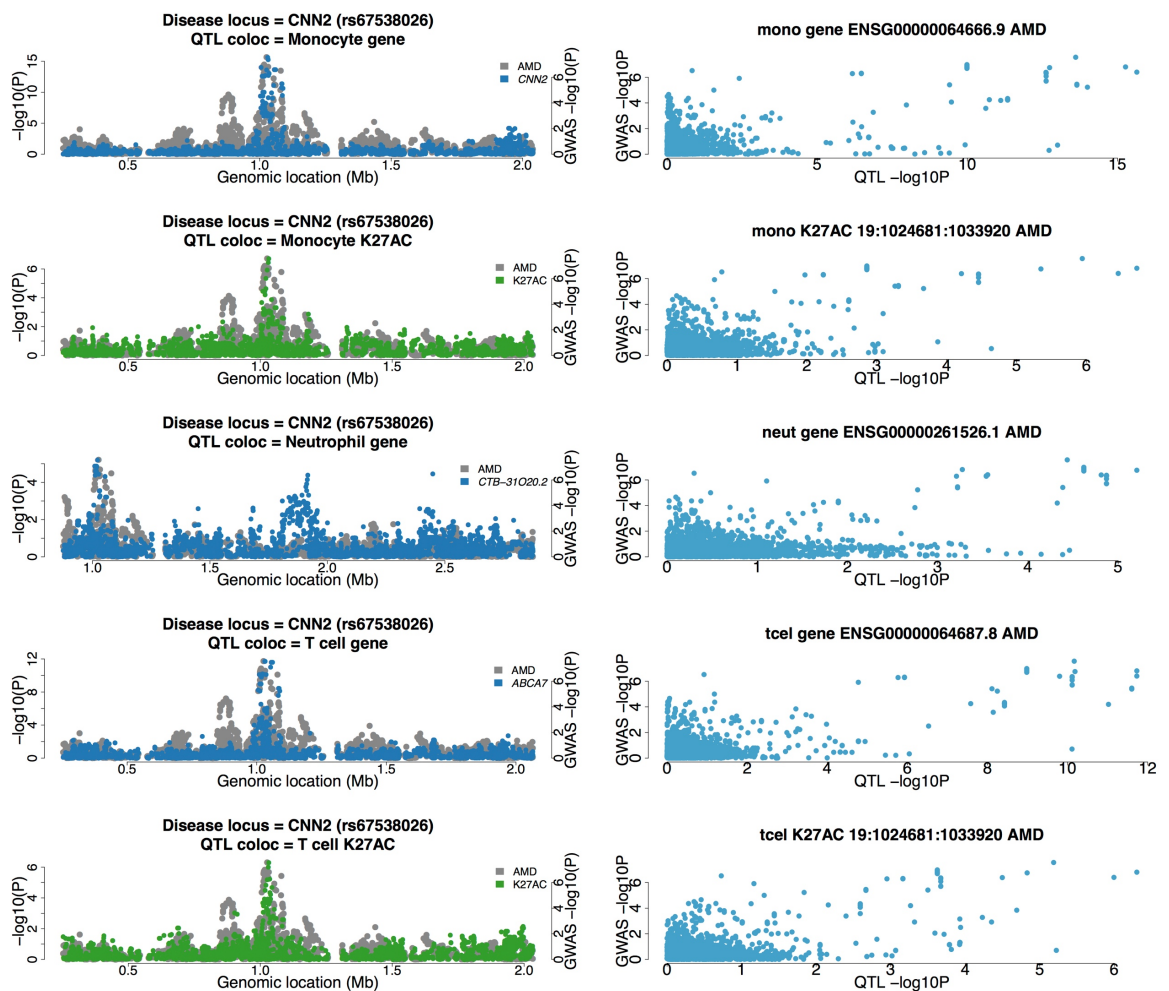


Supplementary Figure 2.2: Changes in gene expression as a result of HL60 gene expression Barplot shows changes in expression of 10 genes known to be affected by HL60 differentiation. Expression was measured by real-time qPCR and the change in expression is evaluated with respect to HL60 cells grown for 24 hours. Figure produced by Stephen Watt.

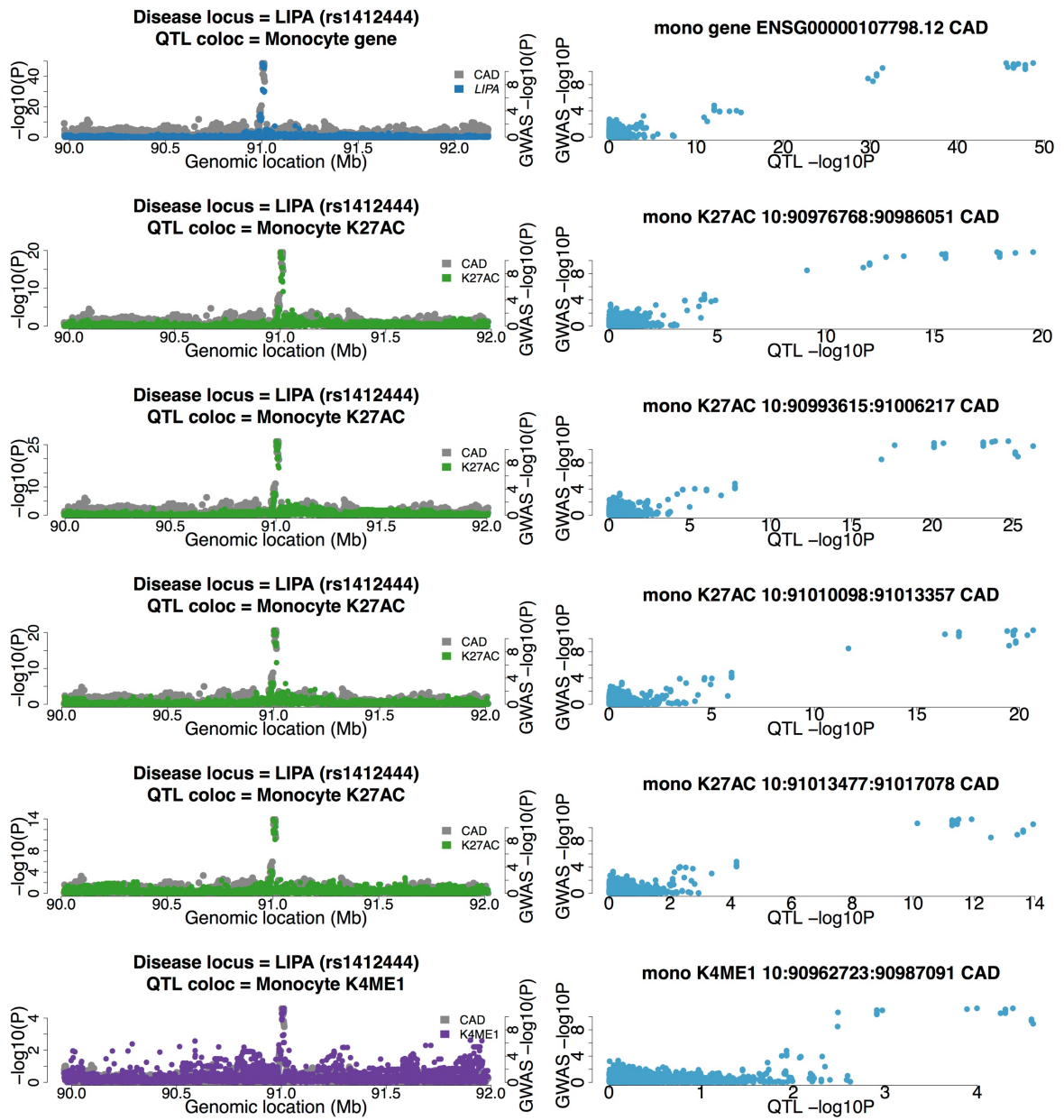
Supplementary Figures 2.3: Examples of disease loci and colocalised molecular features

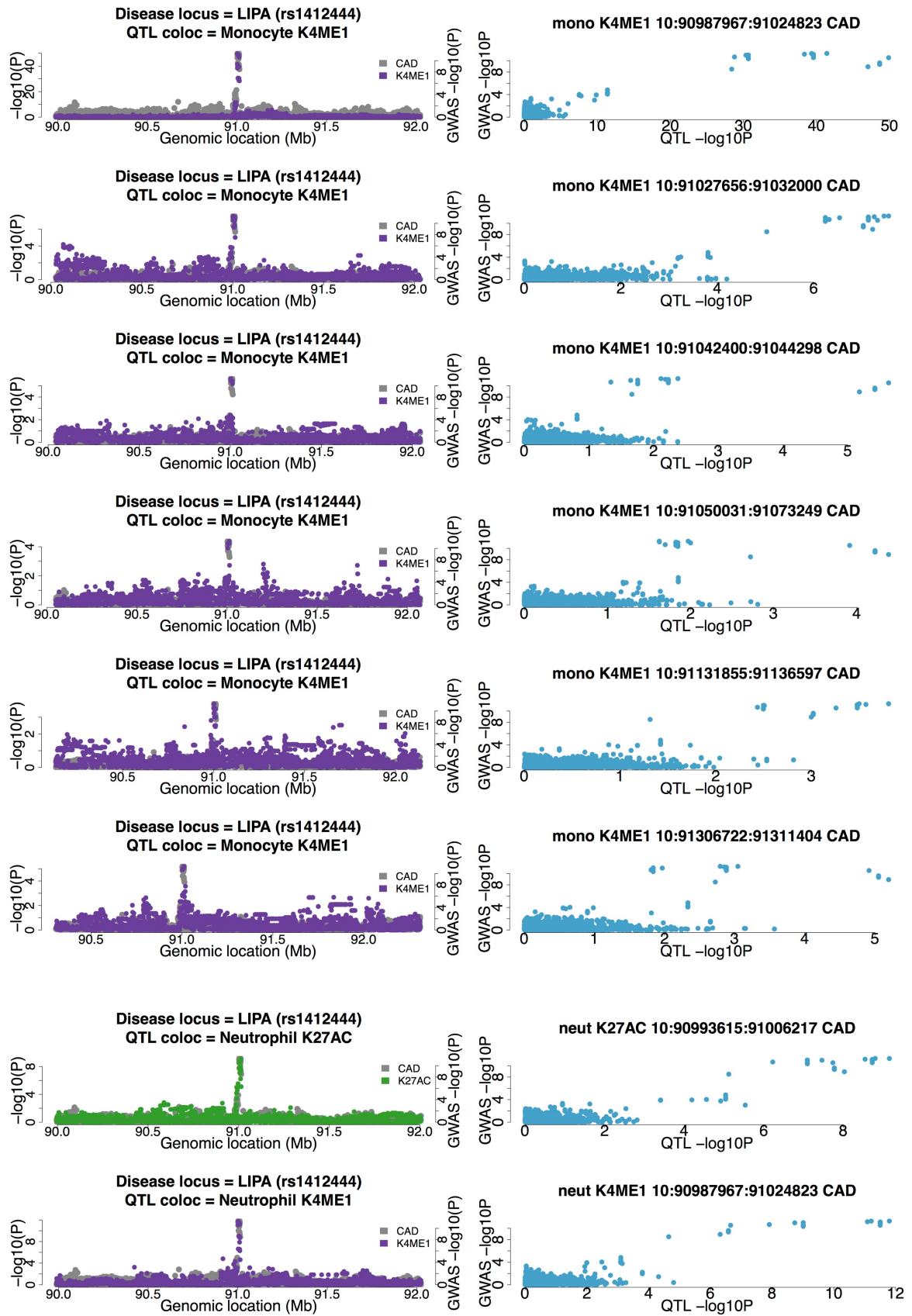
Regional association plots for particular colocalised loci mentioned in the main text of this chapter (left plot). The disease locus as defined by the GWAS study and the lead SNP in brackets is included, as well as the cell type and feature that is colocalised. The eQTL gene is also included in the legend. The correlation between p values of the two traits is also given (right plot). If a locus is colocalised, then the p values in the locus should be correlated. These also provide an initial indication of independent signals that may not be colocalised if the correlation is lower.

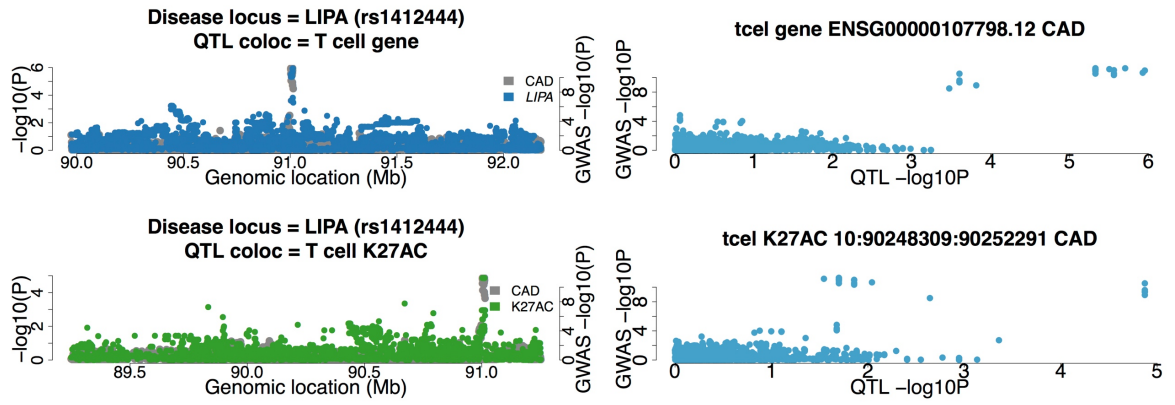
A) Advanced age-related macular degeneration *CNN2* locus



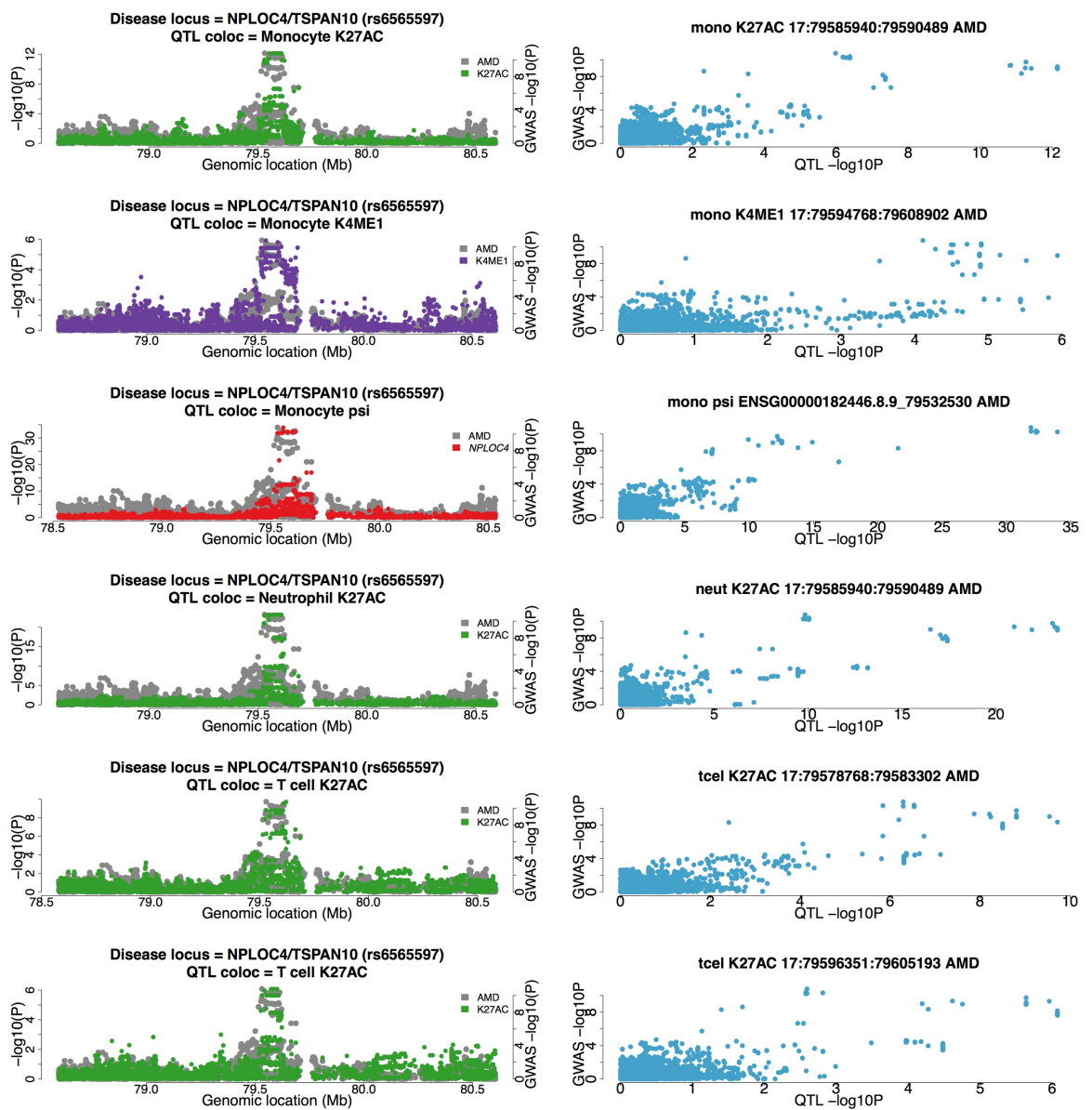
B) Coronary artery disease *LIPA* locus

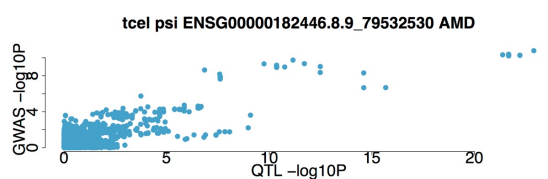
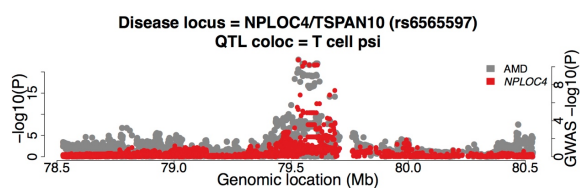
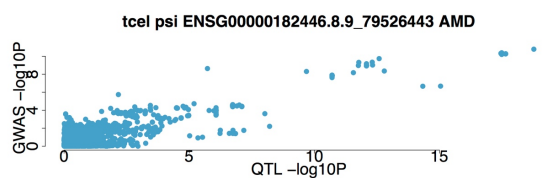
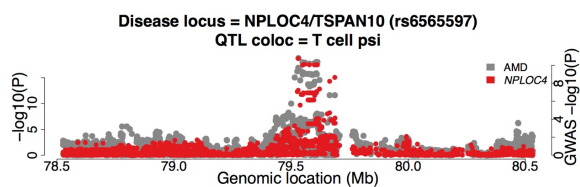
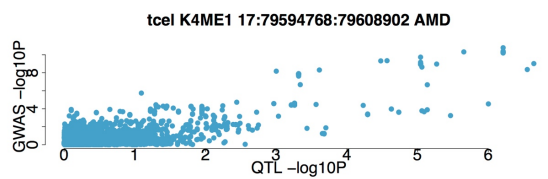
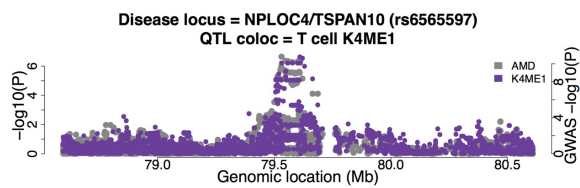
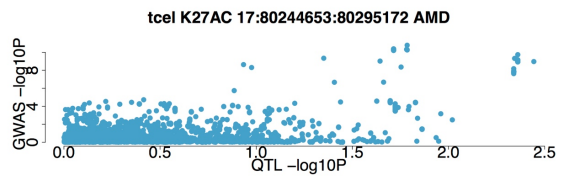
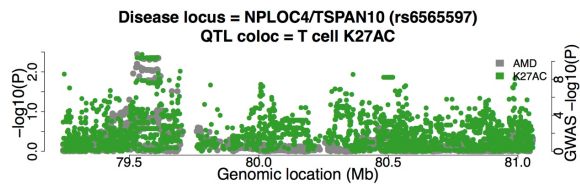
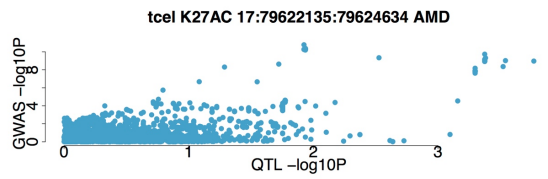
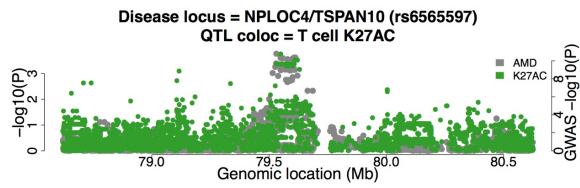
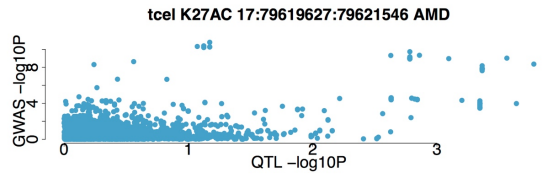
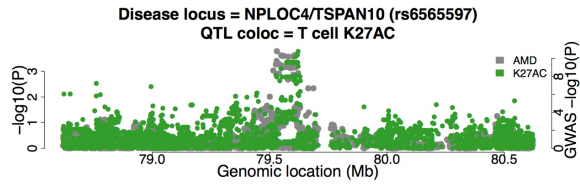




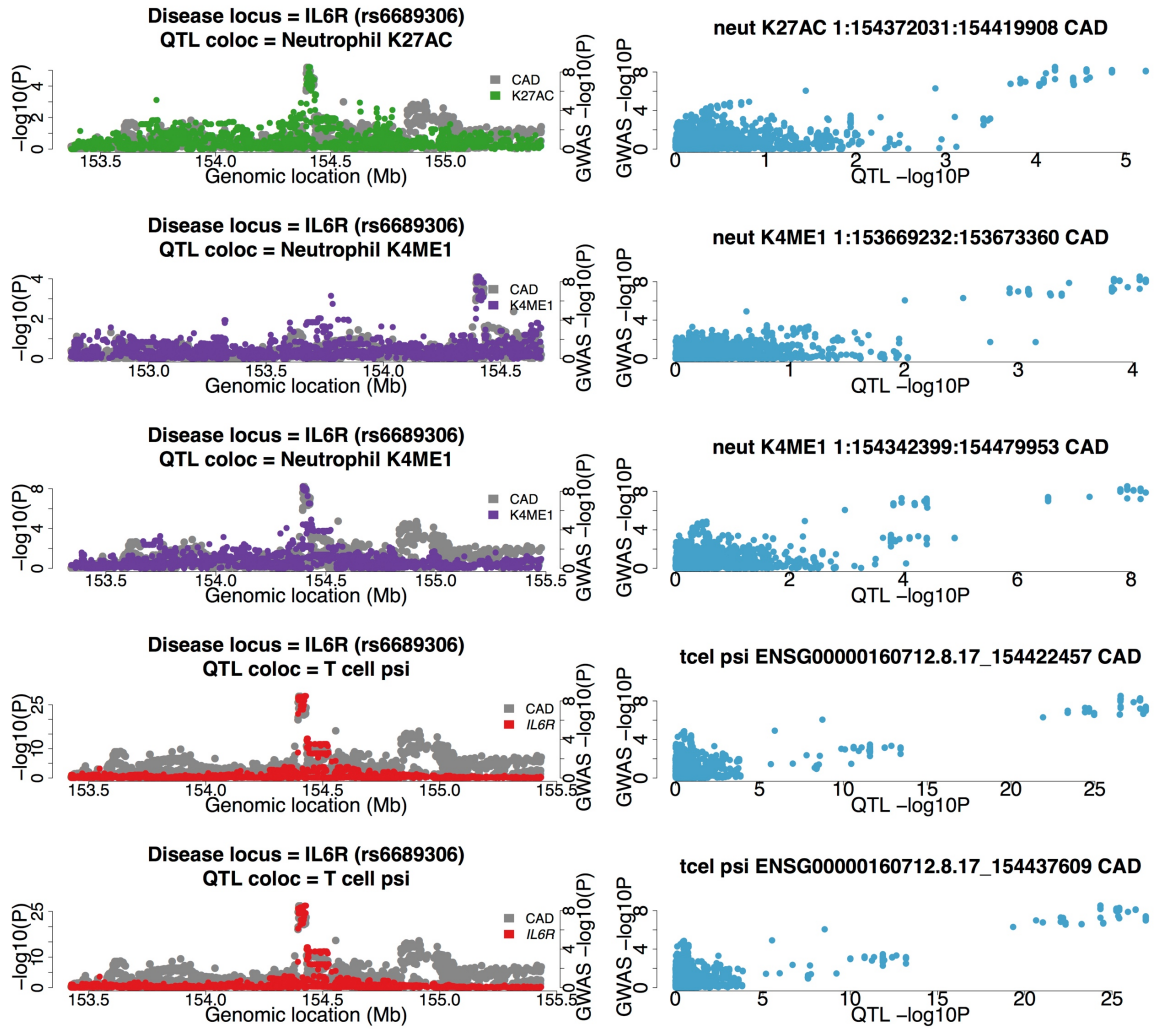


C) Advanced age-related macular degeneration *NPLOC4* locus

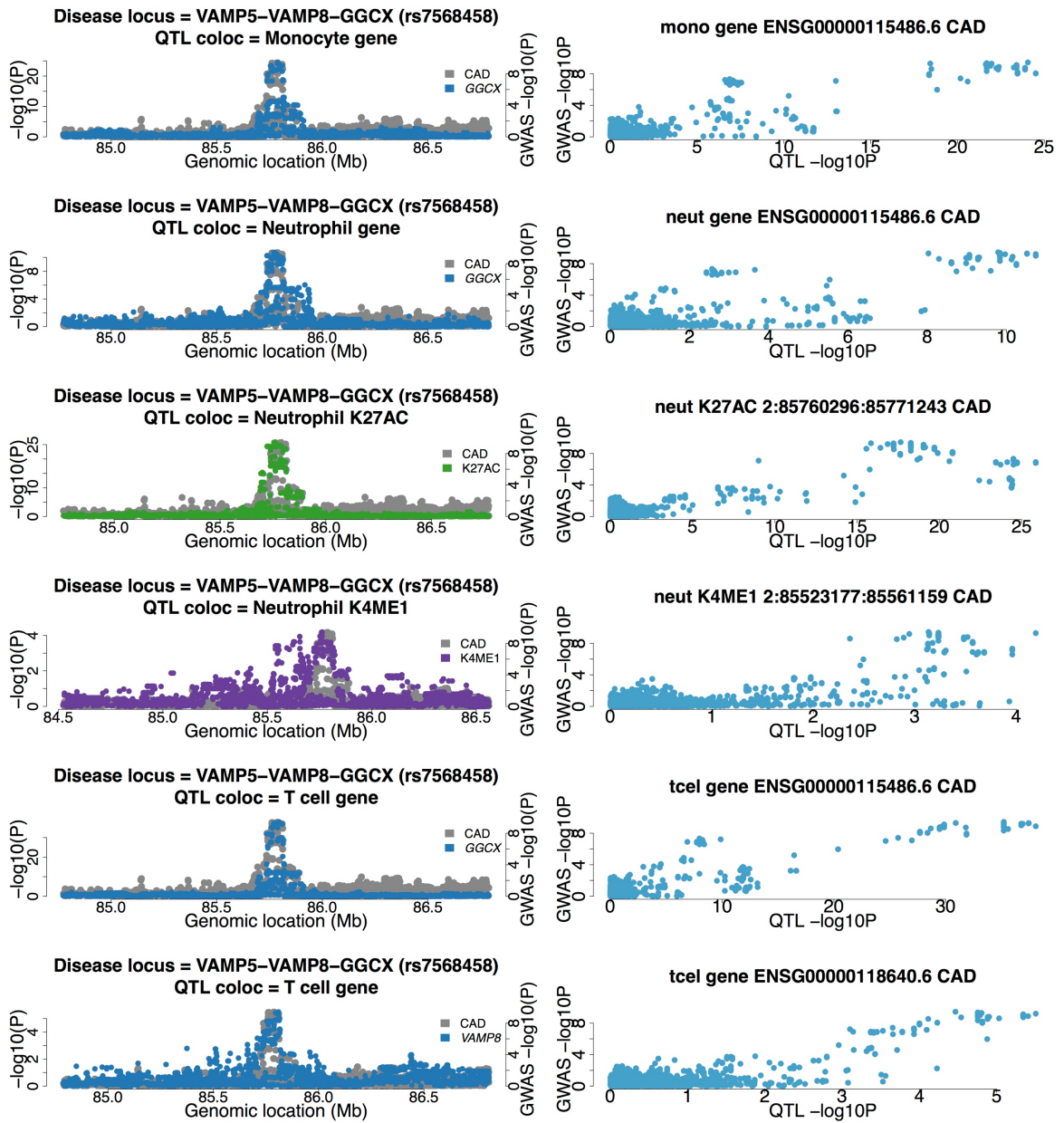




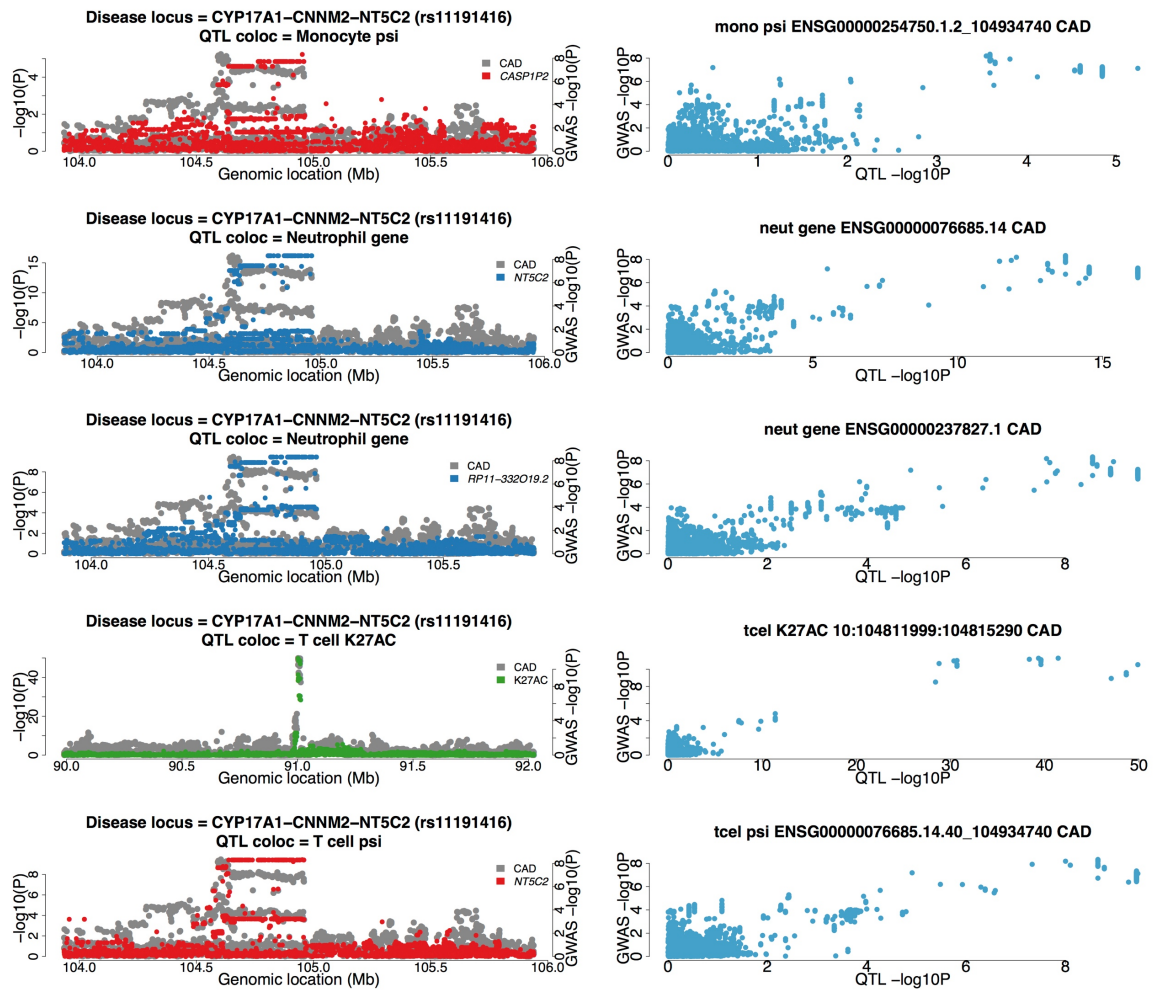
D) Coronary artery disease *IL6R* locus



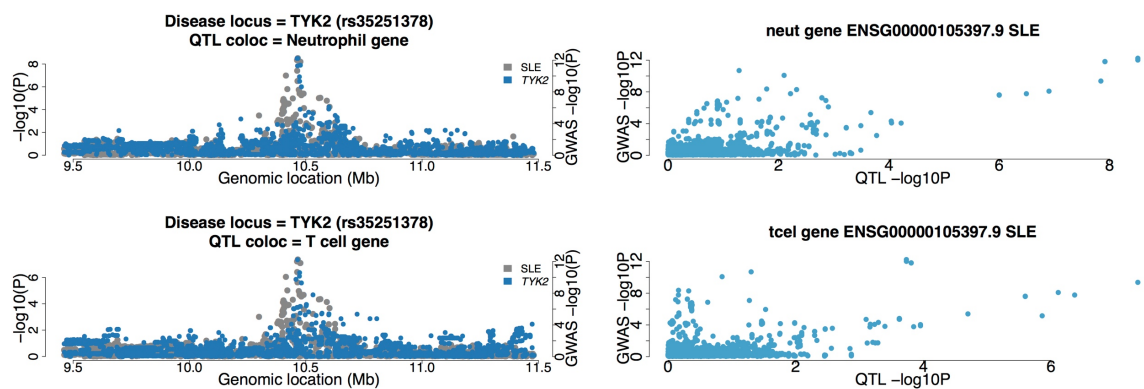
E) Coronary artery disease *VAMP5-VAMP8-GGCX* locus



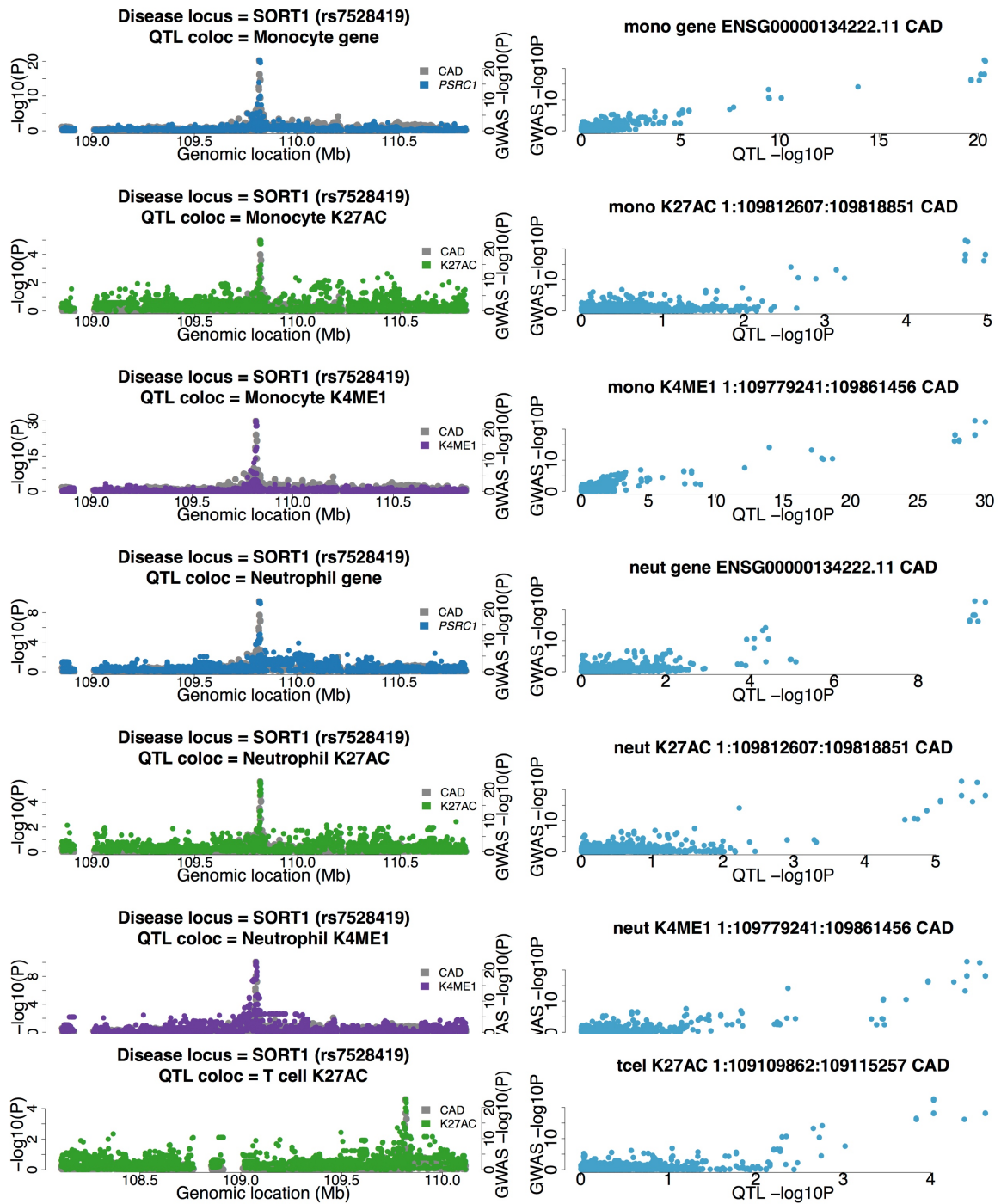
F) Coronary artery disease *CYP17A1-CNNM2-NT5C2* locus



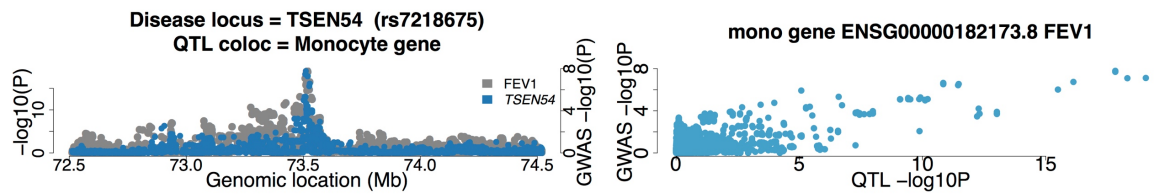
G) Systemic lupus erythematosus *TYK* locus



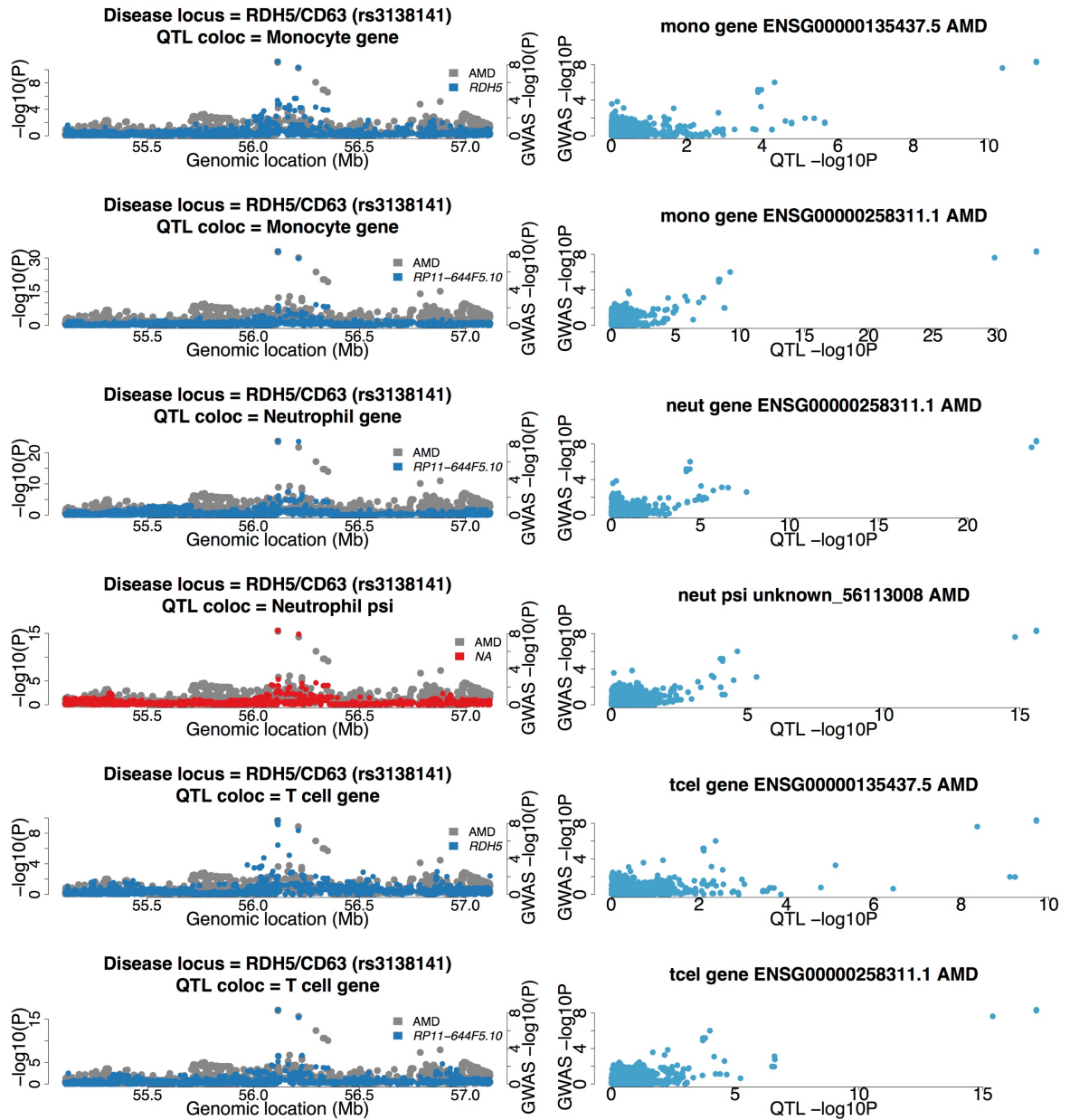
H) Coronary artery disease *SORT1* locus



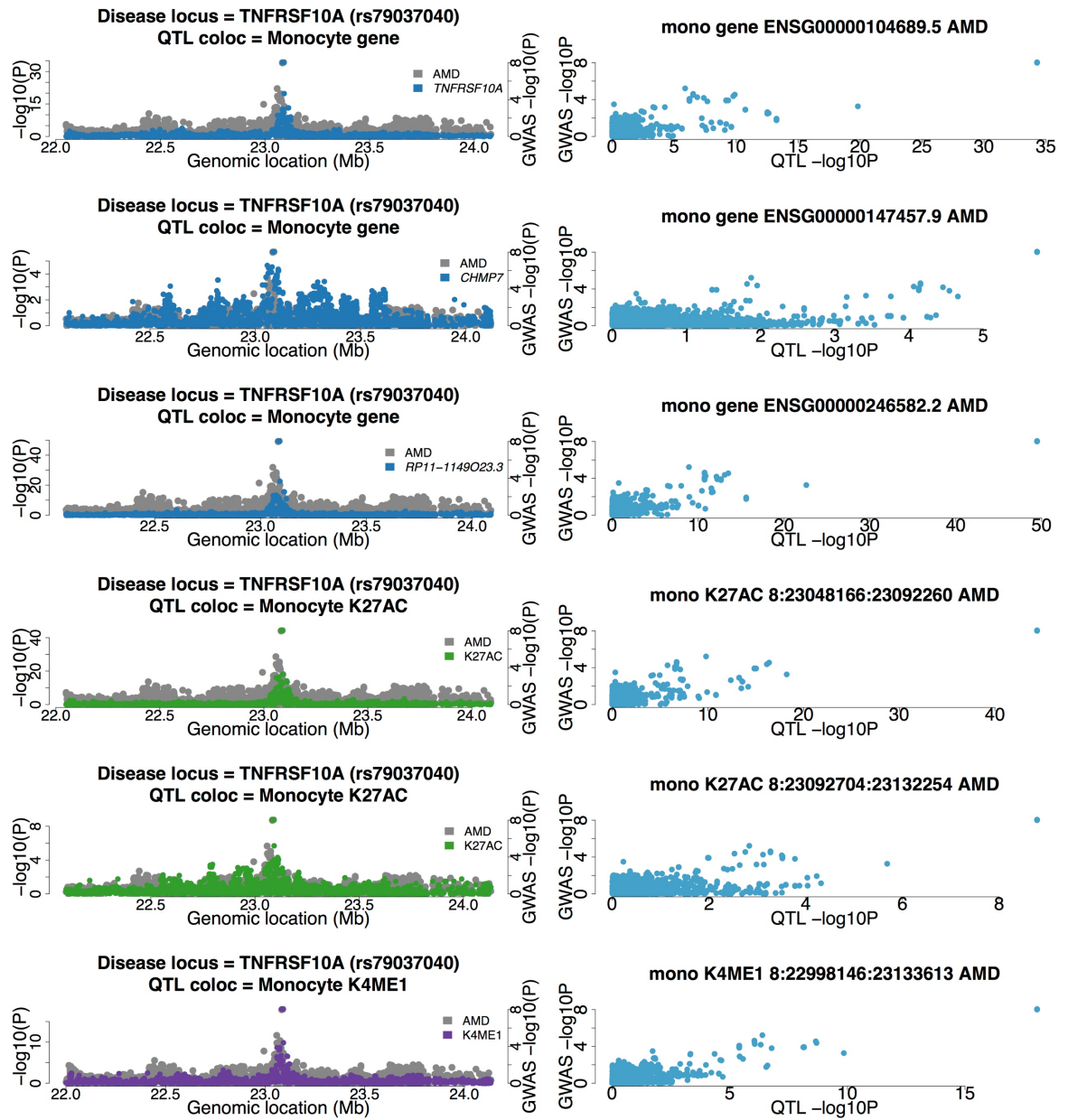
I) Extremes of FEV₁ lung ratio *TSEN54* locus



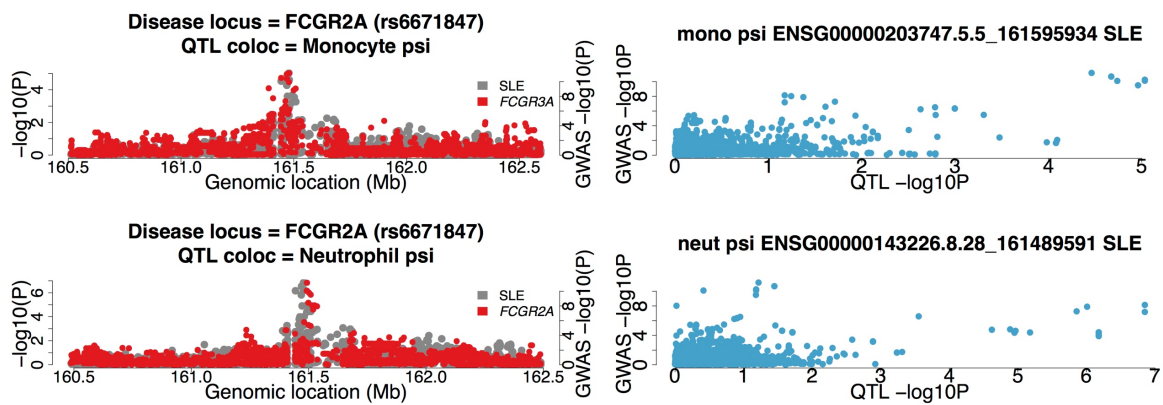
J) Advanced age-related macular degeneration *RDH5* locus



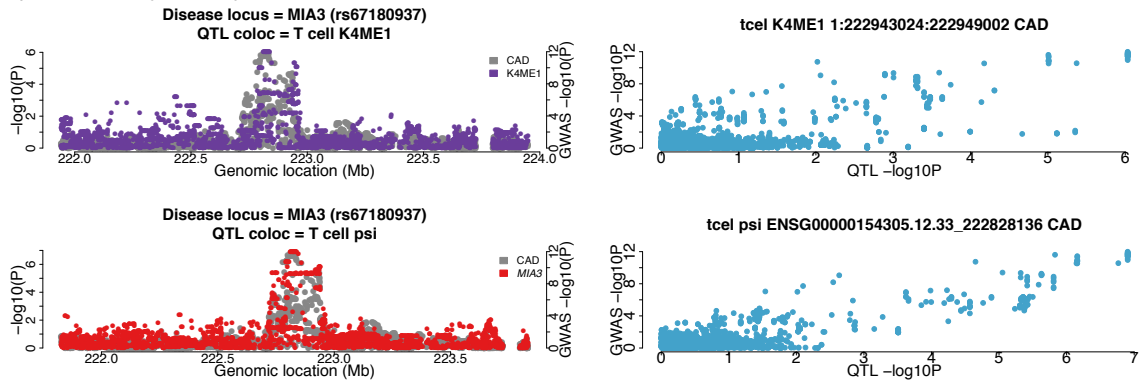
K) Advanced age-related macular degeneration *TNFRSF10A* locus



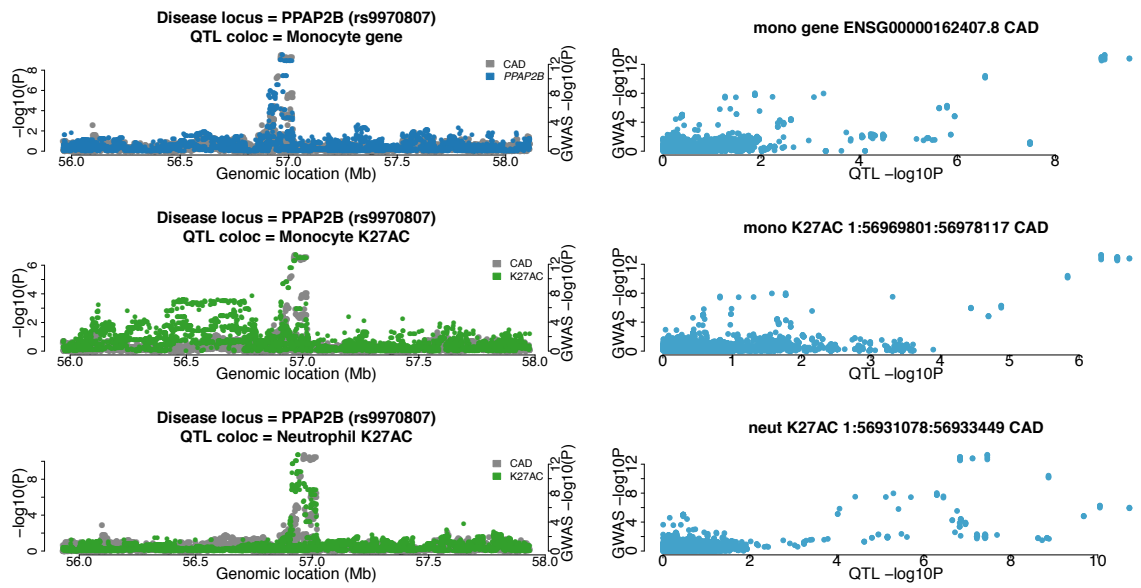
L) Systemic lupus erythematosus *FCGR2A* locus



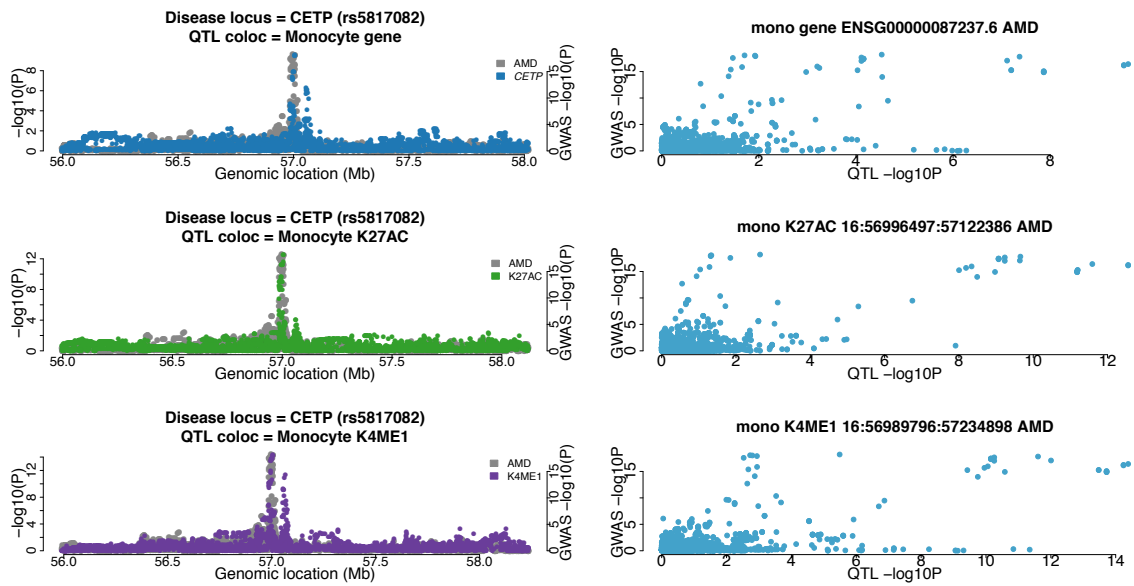
M) Coronary artery disease *MIA3* locus



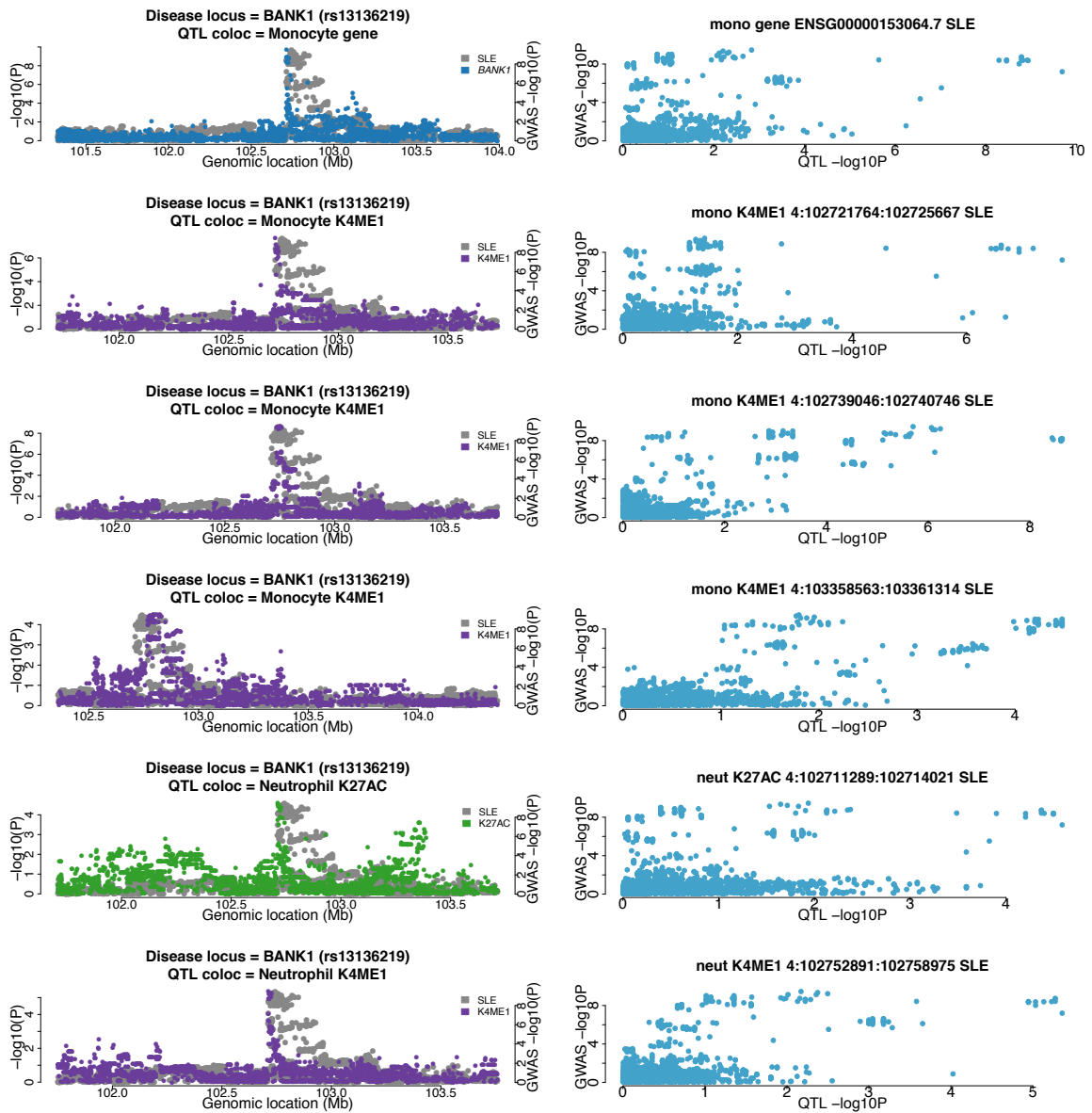
N) Coronary artery disease *PPAP2B* locus



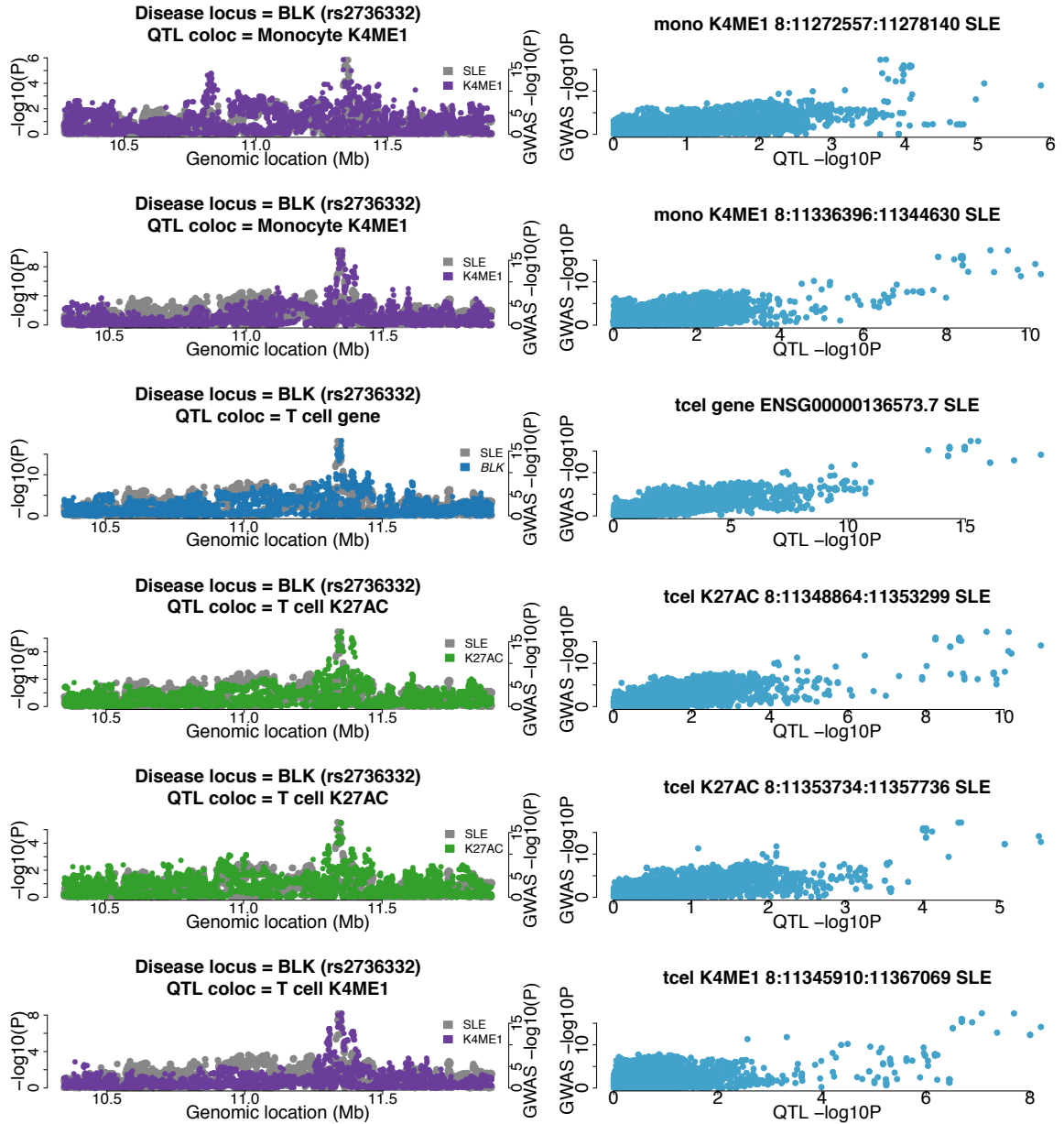
O) Advanced age-related macular degeneration *CETP* locus



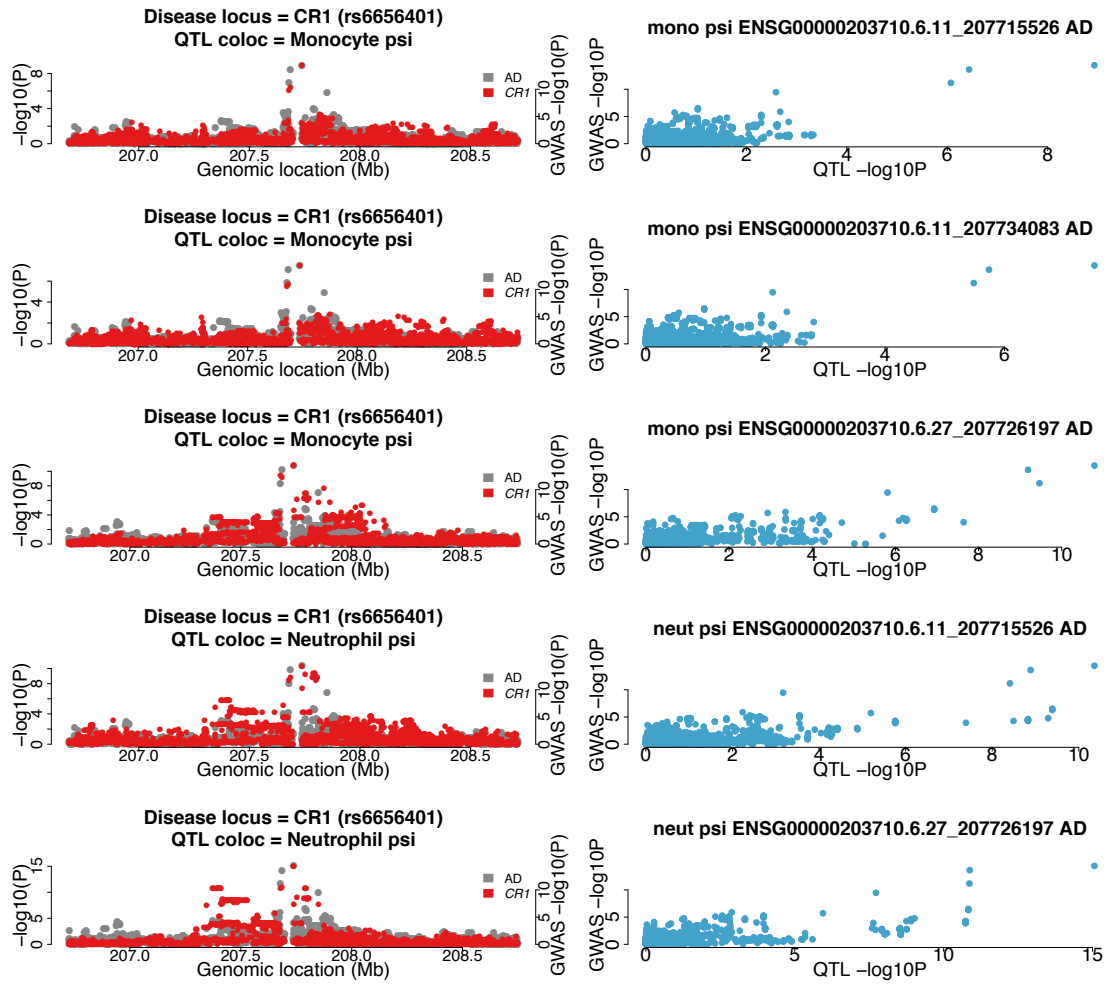
P) Systemic lupus erythematosus *BANK1* locus



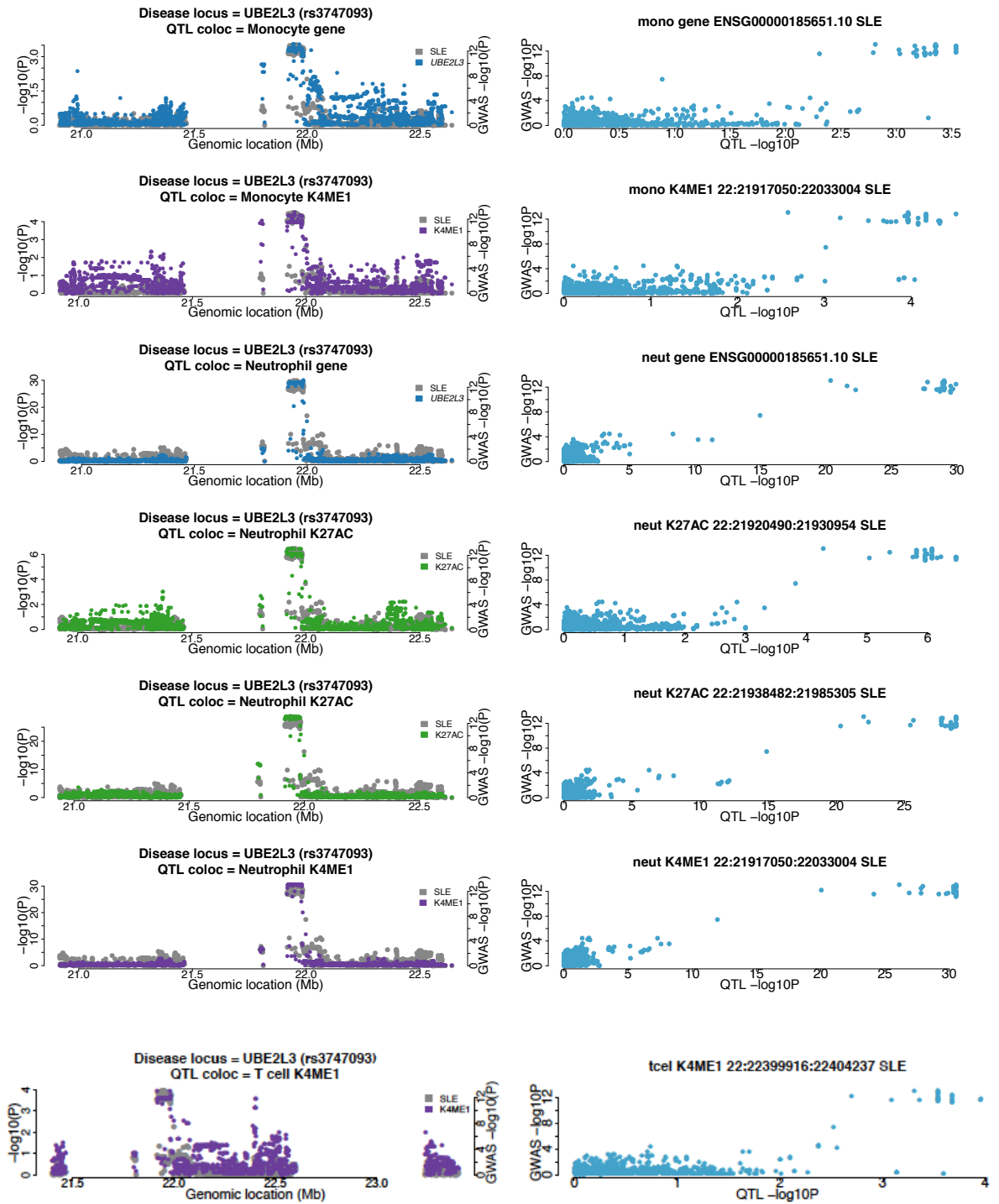
Q) Systemic lupus erythematosus *BLK* locus



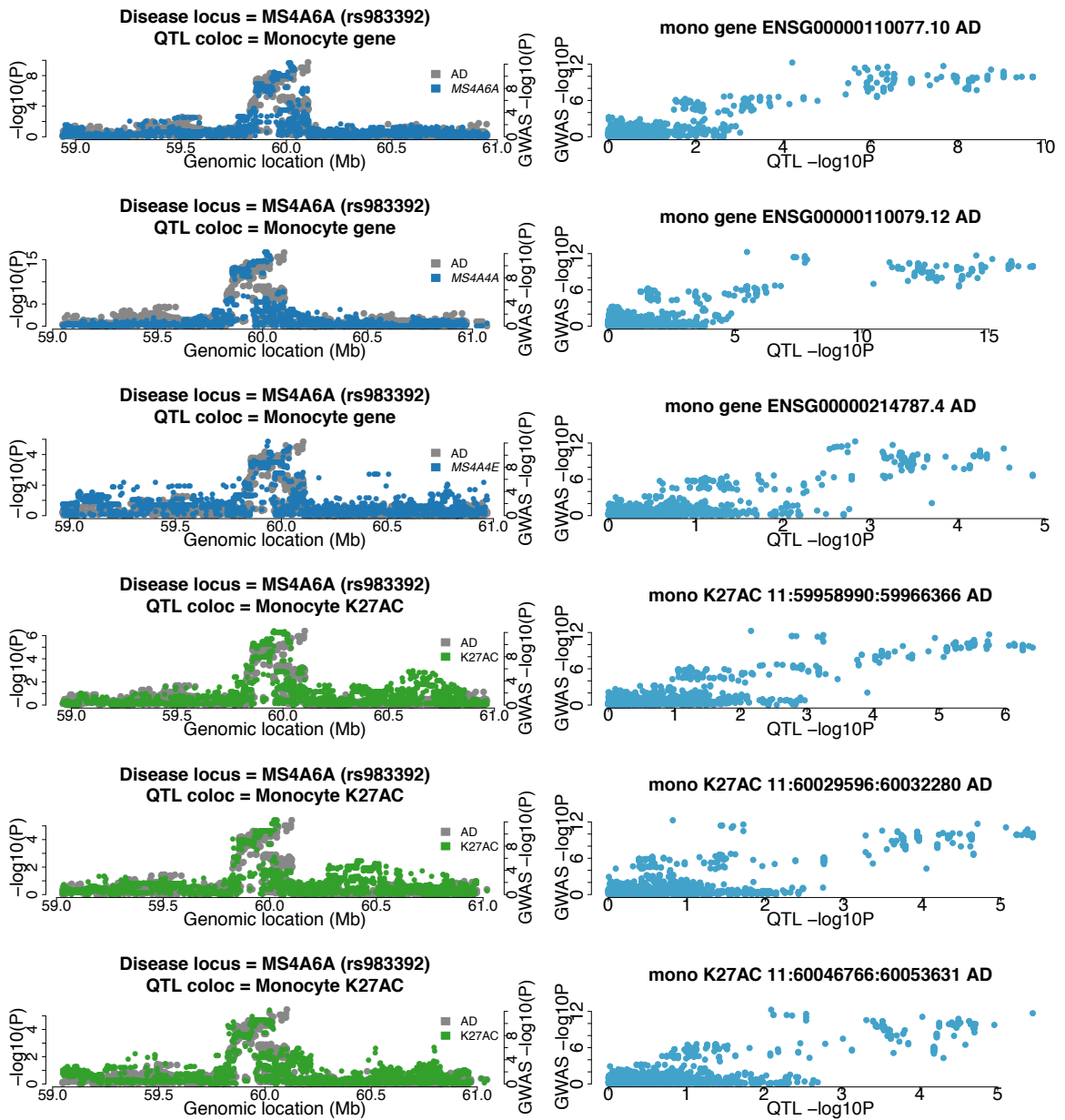
R) Alzheimer's disease *CR1* locus

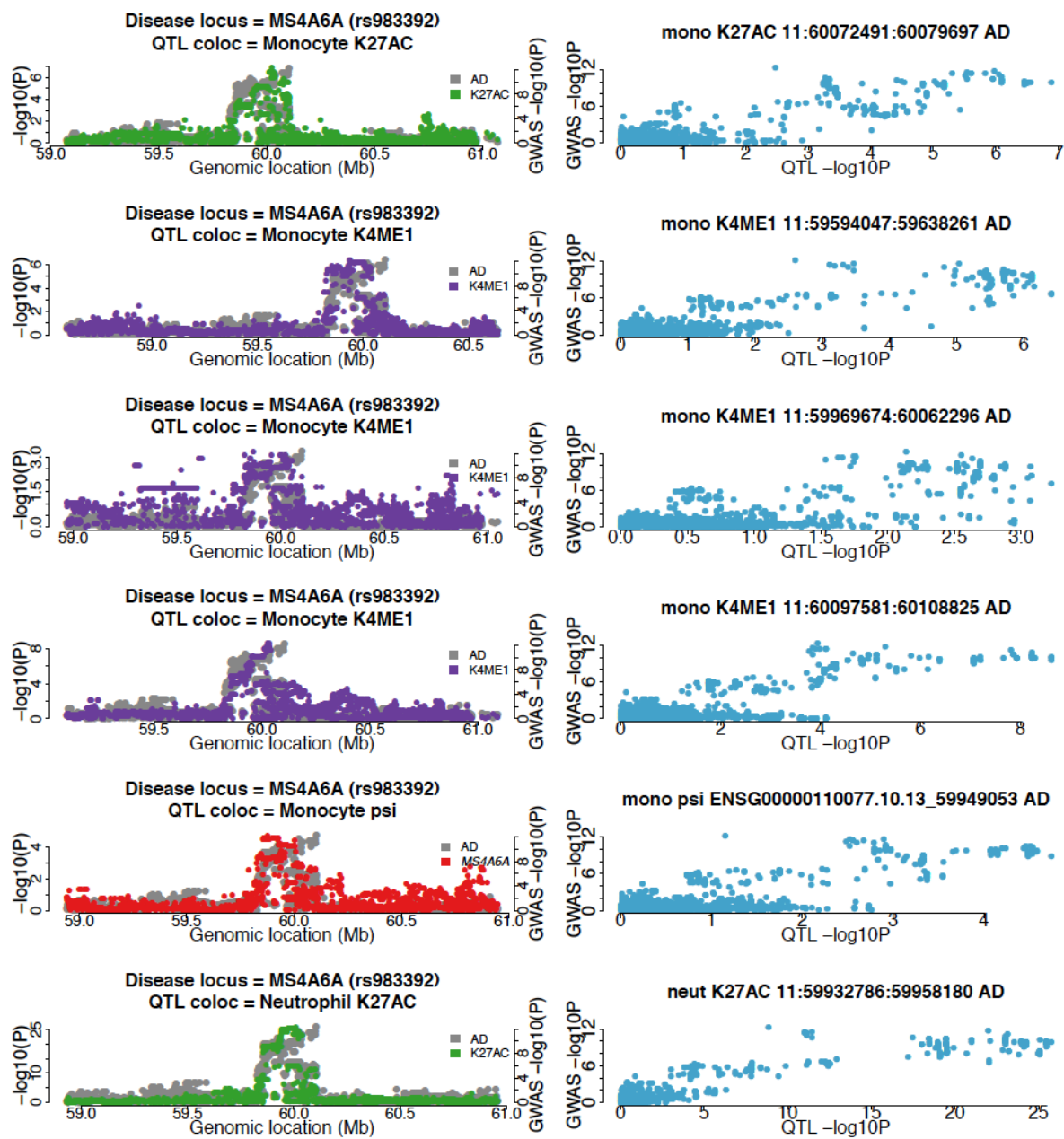


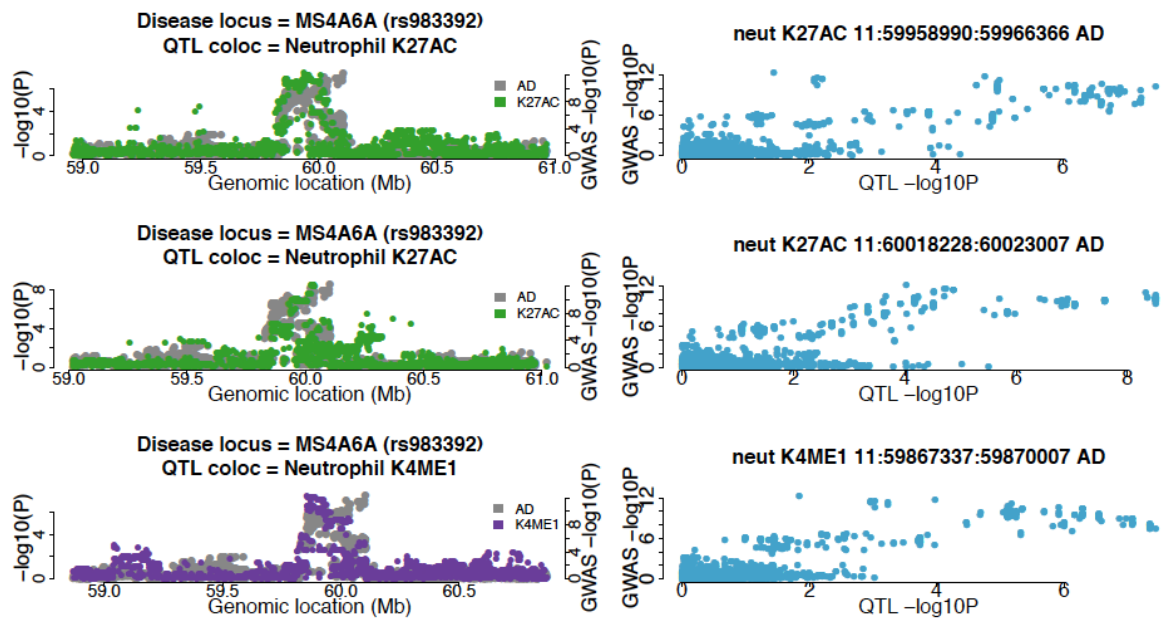
S) Systemic lupus erythematosus *UBE2L3* locus



T) Alzheimer's disease *MS4A6A* locus







Chapter 2 Supplementary Tables

| Feature (SNP) | Model (covariate) | Beta (SE) | P |
|--------------------------------------|--|----------------|---------------------------|
| <i>TNFRSF10A</i> (rs13255394) | Univariate | -0.266 (0.017) | 9.725 x 10 ⁻³⁵ |
| | Conditional (<i>RP11-114O23.3</i> expression) | -0.075 (0.028) | 8.638 x 10 ⁻⁰³ |
| | Conditional (<i>RP11-114O23.3</i> expression + H3K27ac signal) | -0.045 (0.031) | 1.355 x 10 ⁻⁰¹ |
| <i>RP11-114O23.3</i> (rs13255394) | Univariate | -0.880 (0.040) | 5.433 x 10 ⁻⁵⁴ |
| | Conditional (<i>TNFRSF10A</i> expression) | -0.576 (0.052) | 2.312 x 10 ⁻²² |
| | Conditional (<i>TNFRSF10A</i> expression + H3K27ac signal) | -0.565 (0.061) | 2.325 x 10 ⁻¹⁶ |

Supplementary Table 2.1: Conditional causality analysis in the *TNFRSF10A* locus using Blueprint phase 1 genetic data

Association results (beta, SE, and p value) from a simple linear regression model. The univariate approach tests for association of the respective gene expression with the genotype of rs13255394 (lead monocyte SNP). Conditional analysis then tests for association of the gene with genotype whilst conditioning on the expression of the alternative gene. The reduction in p value is greatest when testing for association between the SNP and *TNFRSF10A* expression whilst conditioning on *RP11-114O23.3* expression, which suggests the RNA may be causal for variation in expression of *TNFRSF10A*. The further approach conditions on the gene expression and H3K27ac level.

| | Disease SNP | Study SNP | LD | Locus | Feature SNP | Cell | Mark | Feature.ID |
|----|-------------|------------|----|--------|-------------|------|------|---|
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | M | G | ENSG00000185899.1 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs112524998 | M | G | ENSG00000229153.1 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | M | G | ENSG00000221855.1 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs10265814 | M | K27 | 7:143173365:143179545 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs10237465 | M | K27 | 7:143118095:143120257 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | M | K27 | 7:143067879:143115356 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs12540656 | M | K27 | 7:143115439:143117746 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs10265814 | M | K27 | 7:143158610:143161683 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs10237465 | M | K27 | 7:143122978:143124643 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs112524998 | M | K27 | 7:143133149:143136359 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs112524998 | M | K27 | 7:143154161:143157641 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs10265814 | M | K27 | 7:143200449:143202615 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs10265814 | M | K27 | 7:143180293:143187266 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | M | K27 | 7:143195718:143199833 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | M | K4 | 7:143052447:143144656 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | M | K4 | 7:143147624:143164247 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs6966814 | N | G | ENSG00000229153.1 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | N | G | ENSG00000185899.1 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs6966814 | N | G | ENSG00000234066.1 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs11771145 | N | G | ENSG00000221855.1 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs10265814 | N | K4 | 7:143052447:143144656 |
| AD | rs11771145 | rs11771145 | NA | EPHA1 | rs112524998 | T | S | 7:143104980:143112160:1:3:1, 7:143104984:143112160:1:1:1, 7:143111539:143112160:1:1:0 |
| AD | rs6656401 | rs6656401 | NA | CR1 | rs7515905 | M | S | 1:207685000:207715526:1:1:1, 1:207713412:207715526:1:1:1 |
| AD | rs6656401 | rs6656401 | NA | CR1 | rs7515905 | M | S | 1:207726197:207731882:1:1:1, 1:207726197:207748939:1:1:1 |
| AD | rs6656401 | rs6656401 | NA | CR1 | rs7515905 | M | S | 1:207685000:207734083:1:1:1, 1:207713412:207734083:1:1:1, 1:207731969:207734083:1:1:1 |
| AD | rs6656401 | rs6656401 | NA | CR1 | rs7515905 | N | S | 1:207726197:207731882:1:1:1, 1:207726197:207748939:1:1:1 |
| AD | rs6656401 | rs6656401 | NA | CR1 | rs7515905 | N | S | 1:207685000:207715526:1:1:1, 1:207713412:207715526:1:1:1 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs611418 | M | G | ENSG00000110079.12 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs611418 | M | G | ENSG00000110077.10 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs617135 | M | G | ENSG00000214787.4 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs4938931 | M | K27 | 11:60029596:60032280 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs2081545 | M | K27 | 11:59958990:59966366 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs1562990 | M | K27 | 11:60072491:60079697 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs1582763 | M | K27 | 11:60046766:60053631 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs580064 | M | K4 | 11:59969674:60062296 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs4938931 | M | K4 | 11:60097581:60108825 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs617135 | M | K4 | 11:59594047:59638261 |

| | | | | | | | | |
|-----|-------------|-------------|----|--------|-------------|---|-----|--|
| AD | rs983392 | rs983392 | NA | MS4A6A | rs4939311 | M | S | 11:59943085:59949053:2:2:1, 11:59945790:59949053:2:2:1, 11:59947439:59949053:2:2:1 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs7107627 | N | K27 | 11:60018228:60023007 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs7933202 | N | K27 | 11:59958990:59966366 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs1019671 | N | K27 | 11:59932786:59958180 |
| AD | rs983392 | rs983392 | NA | MS4A6A | rs1441586 | N | K4 | 11:59867337:59870007 |
| AMD | rs10033900 | rs10033900 | NA | CFI | rs3181191 | T | G | ENSG00000248785.1 |
| AMD | rs11080055 | rs11080055 | NA | VTN | rs2027993 | M | G | ENSG00000244045.5 |
| AMD | rs11080055 | rs11080055 | NA | VTN | rs11080055 | N | G | ENSG00000004139.9 |
| AMD | rs11080055 | rs11080055 | NA | VTN | rs241777 | N | G | ENSG00000244045.5 |
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| AMD | rs1142 | rs1142 | NA | SRPK2 | rs12534381 | M | G | ENSG00000135250.12 |
| AMD | rs1142 | rs1142 | NA | SRPK2 | rs10263499 | M | K27 | 7:104849291:104858777 |
| AMD | rs1142 | rs1142 | NA | SRPK2 | rs55671517 | M | K27 | 7:104840590:104849222 |
| AMD | rs1142 | rs1142 | NA | SRPK2 | rs2299304 | M | K27 | 7:104578443:104588833 |
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| AMD | rs140647181 | rs140647181 | NA | COL8A1 | rs6791887 | M | G | ENSG00000036054.8 |
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| AMD | rs3138141 | rs3138141 | NA | RDH5 | rs3138142 | M | G | ENSG00000135437.5 |
| AMD | rs3138141 | rs3138141 | NA | RDH5 | rs3138142 | M | G | ENSG00000258311.1 |
| AMD | rs3138141 | rs3138141 | NA | RDH5 | rs3138142 | N | G | ENSG00000258311.1 |
| AMD | rs3138141 | rs3138141 | NA | RDH5 | rs3138142 | N | S | 12:56113008:56113282:1:1:1, 12:56113008:56115472:1:1:1 |
| AMD | rs3138141 | rs3138141 | NA | RDH5 | rs3138142 | T | G | ENSG00000135437.5 |
| AMD | rs3138141 | rs3138141 | NA | RDH5 | rs3138142 | T | G | ENSG00000258311.1 |
| AMD | rs5817082 | rs5817082 | NA | CETP | rs7205804 | M | G | ENSG00000087237.6 |
| AMD | rs5817082 | rs5817082 | NA | CETP | rs1532625 | M | K27 | 16:56996497:57122386 |
| AMD | rs5817082 | rs5817082 | NA | CETP | rs7205804 | M | K4 | 16:56989796:57234898 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs112612275 | M | K27 | 17:79585940:79590489 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs35816741 | M | K4 | 17:79594768:79608902 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs67050149 | M | S | 17:79526443:79532530:2:2:1, 17:79531006:79532530:2:2:1 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs112612275 | N | K27 | 17:79585940:79590489 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs11655377 | T | K27 | 17:79596351:79605193 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs11150803 | T | K27 | 17:79619627:79621546 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs35816741 | T | K27 | 17:79622135:79624634 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs11150803 | T | K27 | 17:79578768:79583302 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs35816741 | T | K27 | 17:80244653:80295172 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs9912071 | T | K4 | 17:79594768:79608902 |

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|-----|------------|------------|----|---------|-------------|---|-----|--|
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs8070929 | T | S | 17:79526443:79532530:2:2:1, 17:79531006:79532530:2:2:1 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs8070929 | T | S | 17:79526443:79530946:2:2:1, 17:79526443:79532530:2:2:1 |
| AMD | rs6565597 | rs6565597 | NA | NPLOC4 | rs8070929 | T | S | 17:79526443:79527746:2:2:1, 17:79526443:79530946:2:2:1, 17:79526443:79532530:2:2:1 |
| AMD | rs67538026 | rs67538026 | NA | CNN2 | rs10419072 | M | G | ENSG00000064666.9 |
| AMD | rs67538026 | rs67538026 | NA | CNN2 | rs62131196 | M | K27 | 19:1024681:1033920 |
| AMD | rs67538026 | rs67538026 | NA | CNN2 | rs10417845 | N | G | ENSG00000261526.1 |
| AMD | rs67538026 | rs67538026 | NA | CNN2 | rs62131196 | T | G | ENSG00000064687.8 |
| AMD | rs67538026 | rs67538026 | NA | CNN2 | rs62131196 | T | K27 | 19:1024681:1033920 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | G | ENSG00000121716.12 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | G | ENSG00000066923.12 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs7803454 | M | G | ENSG00000146834.8 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | K4 | 7:99906073:99912427 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs113387325 | M | K4 | 7:99801401:99888538 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | S | 7:99954456:99955842:0:0:1, 7:99954507:99955842:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | S | 7:99943592:99947339:1:1:1, 7:99943592:99947353:1:1:1, 7:99943592:99947421:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | S | 7:99954456:99955842:0:0:1, 7:99954507:99955842:1:1:1, 7:99954562:99955842:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | S | 7:99933844:99935801:1:1:1, 7:99933961:99935801:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | S | 7:99943592:99947339:1:1:1, 7:99943592:99947421:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | M | S | 7:99943592:99947339:1:1:1, 7:99943592:99947353:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs67471932 | N | G | ENSG00000233389.2 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | N | G | ENSG00000066923.12 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs7787825 | N | G | ENSG00000146834.8 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | N | K4 | 7:99906073:99912427 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | N | K4 | 7:99801401:99888538 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs67471932 | N | K4 | 7:99912739:99917535 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | N | S | 7:99954456:99955842:0:0:1, 7:99954507:99955842:1:1:1, 7:99954562:99955842:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | N | S | 7:99933844:99935801:1:1:1, 7:99933961:99935801:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | N | S | 7:99954456:99955842:0:0:1, 7:99954507:99955842:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs11769886 | N | S | 7:99943592:99947339:1:1:1, 7:99943592:99947421:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs11769886 | N | S | 7:99943592:99947339:1:1:1, 7:99943592:99947353:1:1:1, 7:99943592:99947421:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs11769886 | N | S | 7:99943592:99947339:1:1:1, 7:99943592:99947353:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | T | G | ENSG00000085514.10 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | T | G | ENSG00000078487.13 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs113387325 | T | G | ENSG00000241357.1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs113387325 | T | G | ENSG00000121716.12 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs7787825 | T | G | ENSG00000146834.8 |

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|-----|----------------------|------------------|----|---------------|-------------|---|-----|--|
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | T | S | 7:99951107:99951517:1:1:1, 7:99951107:99952765:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs113387325 | T | S | 7:99933844:99935801:1:1:1, 7:99933961:99935801:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs113387325 | T | S | 7:99947511:99949785:1:1:0, 7:99949034:99949785:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | T | S | 7:99943592:99947339:1:1:1, 7:99943592:99947353:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs113387325 | T | S | 7:99947511:99948874:1:1:1, 7:99947511:99949785:1:1:0 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | T | S | 7:99943592:99947339:1:1:1, 7:99943592:99947421:1:1:1 |
| AMD | rs7803454 | rs7803454 | NA | PILRB/A | rs111312383 | T | S | 7:99943592:99947339:1:1:1, 7:99943592:99947353:1:1:1, 7:99943592:99947421:1:1:1 |
| AMD | rs79037040 | rs79037040 | NA | TNFRSF 10A | rs13255394 | M | G | ENSG00000246582.2 |
| AMD | rs79037040 | rs79037040 | NA | TNFRSF 10A | rs13255394 | M | G | ENSG00000147457.9 |
| AMD | rs79037040 | rs79037040 | NA | TNFRSF 10A | rs13255394 | M | G | ENSG00000104689.5 |
| AMD | rs79037040 | rs79037040 | NA | TNFRSF 10A | rs13255394 | M | K27 | 8:23048166:23092260 |
| AMD | rs79037040 | rs79037040 | NA | TNFRSF 10A | rs13255394 | M | K27 | 8:23092704:23132254 |
| AMD | rs79037040 | rs79037040 | NA | TNFRSF 10A | rs13255394 | M | K4 | 8:22998146:23133613 |
| CAD | chr2:203828796 :l | chr2:203828796:l | NA | WDR12 | rs148513392 | M | G | ENSG00000163596.12 |
| CAD | chr2:203828796 :l | chr2:203828796:l | NA | WDR12 | rs78488377 | M | G | ENSG00000144426.14 |
| CAD | chr2:203828796 :l | chr2:203828796:l | NA | WDR12 | rs140201293 | M | G | ENSG00000138380.12 |
| CAD | chr2:203828796 :l | chr2:203828796:l | NA | WDR12 | rs16839813 | M | K4 | 2:204391511:204403180 |
| CAD | chr2:203828796 :l | chr2:203828796:l | NA | WDR12 | rs4675290 | N | K27 | 2:204364327:204367436 |
| CAD | chr2:203828796 :l | chr2:203828796:l | NA | WDR12 | rs72934535 | N | K27 | 2:203772046:203778875 |
| CAD | chr2:203828796 :l | chr2:203828796:l | NA | WDR12 | rs72932553 | T | G | ENSG00000236047.1 |
| CAD | rs11191416 | rs11191416 | NA | NT5C2 | rs79780963 | M | S | 10:104934740:104940941:2:2:1, 10:104934740:104940987:2:2:0 |
| CAD | rs11191416 | rs11191416 | NA | NT5C2 | rs111374294 | N | G | ENSG00000237827.1 |
| CAD | rs11191416 | rs11191416 | NA | NT5C2 | rs111374294 | N | G | ENSG00000076685.14 |
| CAD | rs11191416 | rs11191416 | NA | NT5C2 | rs3740390 | T | K27 | 10:104811999:104815290 |
| CAD | rs11191416 | rs11191416 | NA | NT5C2 | rs79780963 | T | S | 10:104934740:104936242:2:2:0, 10:104934740:104937274:2:2:0, 10:104934740:104940941:2:2:1, 10:104934740:104940987:2:2:0, 10:104934740:104952992:2:2:1 |

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|-----|------------|------------|----|-------|------------|---|-----|--|
| CAD | rs11191416 | rs11191416 | NA | NT5C2 | rs79780963 | T | S | 10:104934740:104940941:2:2:1, 10:104934740:104940987:2:2:0, 10:104934740:104952992:2:2:1 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1412444 | M | G | ENSG00000107798.12 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1320496 | M | K27 | 10:90993615:91006217 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1412444 | M | K27 | 10:90976768:90986051 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1412444 | M | K27 | 10:91010098:91013357 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1320496 | M | K27 | 10:91013477:91017078 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1332326 | M | K4 | 10:91050031:91073249 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1332326 | M | K4 | 10:91306722:91311404 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1412444 | M | K4 | 10:91131855:91136597 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1320496 | M | K4 | 10:90987967:91024823 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1320496 | M | K4 | 10:91042400:91044298 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1332326 | M | K4 | 10:90962723:90987091 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1412444 | M | K4 | 10:91027656:91032000 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1332328 | N | K27 | 10:90993615:91006217 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1412444 | N | K4 | 10:90987967:91024823 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1332327 | T | G | ENSG00000107798.12 |
| CAD | rs1412444 | rs1412444 | NA | LIPA | rs1320496 | T | K27 | 10:90248309:90252291 |
| CAD | rs17087335 | rs17087335 | NA | REST | rs12645070 | M | G | ENSG00000084093.11 |
| CAD | rs17087335 | rs17087335 | NA | REST | rs12645070 | M | K27 | 4:57771837:57788561 |
| CAD | rs17087335 | rs17087335 | NA | REST | rs12645070 | M | K27 | 4:57823529:57826313 |
| CAD | rs17087335 | rs17087335 | NA | REST | rs12645070 | M | K4 | 4:57770788:57806908 |
| CAD | rs17087335 | rs17087335 | NA | REST | rs12645070 | M | K4 | 4:57820927:57828891 |
| CAD | rs17087335 | rs17087335 | NA | REST | rs12645070 | N | K27 | 4:57823529:57826313 |
| CAD | rs17087335 | rs17087335 | NA | REST | rs6554401 | N | K4 | 4:57820927:57828891 |
| CAD | rs6689306 | rs6689306 | NA | IL6R | rs7549250 | N | K27 | 1:154372031:154419908 |
| CAD | rs6689306 | rs6689306 | NA | IL6R | rs7549338 | N | K4 | 1:153669232:153673360 |
| CAD | rs6689306 | rs6689306 | NA | IL6R | rs11265611 | N | K4 | 1:154342399:154479953 |
| CAD | rs6689306 | rs6689306 | NA | IL6R | rs4845625 | T | S | 1:154422457:154437609:1:1:1, 1:154427058:154437609:1:1:1 |
| CAD | rs6689306 | rs6689306 | NA | IL6R | rs4845625 | T | S | 1:154422457:154426963:1:1:1, 1:154422457:154437609:1:1:1 |
| CAD | rs6689306 | rs6689306 | NA | IL6R | rs4845625 | T | S | 1:154422457:154426548:1:1:0, 1:154422457:154426963:1:1:1, 1:154422457:154437609:1:1:1 |
| CAD | rs67180937 | rs67180937 | NA | MIA3 | rs35700460 | T | K4 | 1:222943024:222949002 |
| CAD | rs67180937 | rs67180937 | NA | MIA3 | rs35700460 | T | S | 1:222828136:222831263:1:1:0, 1:222828136:222832063:1:1:1 |
| CAD | rs7528419 | rs7528419 | NA | SORT1 | rs12740374 | M | G | ENSG00000134222.11 |
| CAD | rs7528419 | rs7528419 | NA | SORT1 | rs660240 | M | K27 | 1:109812607:109818851 |
| CAD | rs7528419 | rs7528419 | NA | SORT1 | rs12740374 | M | K4 | 1:109779241:109861456 |
| CAD | rs7528419 | rs7528419 | NA | SORT1 | rs12740374 | N | G | ENSG00000134222.11 |
| CAD | rs7528419 | rs7528419 | NA | SORT1 | rs660240 | N | K27 | 1:109812607:109818851 |
| CAD | rs7528419 | rs7528419 | NA | SORT1 | rs660240 | N | K4 | 1:109779241:109861456 |
| CAD | rs7528419 | rs7528419 | NA | SORT1 | rs660240 | T | K27 | 1:109109862:109115257 |

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|------|------------|------------|------|--------|------------|---|-----|-----------------------|
| CAD | rs7568458 | rs7568458 | NA | GGCX | rs12714145 | M | G | ENSG00000115486.6 |
| CAD | rs7568458 | rs7568458 | NA | GGCX | rs1078004 | N | G | ENSG00000115486.6 |
| CAD | rs7568458 | rs7568458 | NA | GGCX | rs56819945 | N | K27 | 2:85760296:85771243 |
| CAD | rs7568458 | rs7568458 | NA | GGCX | rs10176176 | N | K4 | 2:85523177:85561159 |
| CAD | rs7568458 | rs7568458 | NA | GGCX | rs11891260 | T | G | ENSG00000115486.6 |
| CAD | rs7568458 | rs7568458 | NA | GGCX | rs1561198 | T | G | ENSG00000118640.6 |
| CAD | rs9970807 | rs9970807 | NA | PPAP2B | rs56186267 | M | G | ENSG00000162407.8 |
| CAD | rs9970807 | rs9970807 | NA | PPAP2B | rs56186267 | M | K27 | 1:56969801:56978117 |
| CAD | rs9970807 | rs9970807 | NA | PPAP2B | rs6588634 | N | K27 | 1:56931078:56933449 |
| FEV1 | rs7218675 | rs7218675 | NA | TSEN54 | rs35643020 | M | G | ENSG00000182173.8 |
| FEV1 | rs78420228 | rs78420228 | NA | CDC123 | rs12241367 | M | K27 | 10:12310277:12315701 |
| FEV1 | rs78420228 | rs78420228 | NA | CDC123 | rs61848342 | M | K27 | 10:12374604:12376790 |
| FEV1 | rs78420228 | rs78420228 | NA | CDC123 | rs11599700 | M | K4 | 10:12273289:12320006 |
| FEV1 | rs78420228 | rs78420228 | NA | CDC123 | rs10795944 | N | G | ENSG00000228302.2 |
| FEV1 | rs7842022 | rs78420228 | NA | CDC123 | rs12241367 | N | K4 | 10:12273289:12320006 |
| SLE | rs1143679 | rs34572943 | 0.94 | ITGAM | rs34082782 | M | G | ENSG00000140688.12 |
| SLE | rs1143679 | rs34572943 | 0.94 | ITGAM | rs34550882 | N | G | ENSG00000261385.1 |
| SLE | rs1143679 | rs34572943 | 0.94 | ITGAM | rs34550882 | N | G | ENSG00000260219.1 |
| SLE | rs1143679 | rs34572943 | 0.94 | ITGAM | rs9673398 | N | G | ENSG00000169896.11 |
| SLE | rs1143679 | rs34572943 | 0.94 | ITGAM | rs9673404 | T | K4 | 16:31355247:31421179 |
| SLE | rs13136219 | rs10028805 | 0.98 | BANK1 | rs7683892 | M | G | ENSG00000153064.7 |
| SLE | rs13136219 | rs10028805 | 0.98 | BANK1 | rs34029191 | M | K4 | 4:102739046:102740746 |
| SLE | rs13136219 | rs10028805 | 0.98 | BANK1 | rs4270588 | M | K4 | 4:103358563:103361314 |
| SLE | rs13136219 | rs10028805 | 0.98 | BANK1 | rs7683892 | M | K4 | 4:102721764:102725667 |
| SLE | rs13136219 | rs10028805 | 0.98 | BANK1 | rs7683892 | N | K27 | 4:102711289:102714021 |
| SLE | rs13136219 | rs10028805 | 0.98 | BANK1 | rs7683892 | N | K4 | 4:102752891:102758975 |
| SLE | rs2736332 | rs2736340 | 0.9 | BLK | rs13257831 | M | K4 | 8:11336396:11344630 |
| SLE | rs2736332 | rs2736340 | 0.9 | BLK | rs12680762 | M | K4 | 8:11272557:11278140 |
| SLE | rs2736332 | rs2736340 | 0.9 | BLK | rs922483 | T | G | ENSG00000136573.7 |
| SLE | rs2736332 | rs2736340 | 0.9 | BLK | rs2736345 | T | K27 | 8:11353734:11357736 |
| SLE | rs2736332 | rs2736340 | 0.9 | BLK | rs922483 | T | K27 | 8:11348864:11353299 |
| SLE | rs2736332 | rs2736340 | 0.9 | BLK | rs922483 | T | K4 | 8:11345910:11367069 |
| SLE | rs35251378 | rs2304256 | 0.95 | TYK2 | rs11085725 | N | G | ENSG00000105397.9 |
| SLE | rs35251378 | rs2304256 | 0.95 | TYK2 | rs280497 | T | G | ENSG00000105397.9 |
| SLE | rs3747093 | rs7444 | 0.88 | UBE2L3 | rs5749485 | M | G | ENSG00000185651.10 |
| SLE | rs3747093 | rs7444 | 0.88 | UBE2L3 | rs2070512 | M | K4 | 22:21917050:22033004 |
| SLE | rs3747093 | rs7444 | 0.88 | UBE2L3 | rs2298429 | N | G | ENSG00000185651.10 |
| SLE | rs3747093 | rs7444 | 0.88 | UBE2L3 | rs11089620 | N | K27 | 22:21920490:21930954 |
| SLE | rs3747093 | rs7444 | 0.88 | UBE2L3 | rs140488 | N | K27 | 22:21938482:21985305 |
| SLE | rs3747093 | rs7444 | 0.88 | UBE2L3 | rs140488 | N | K4 | 22:21917050:22033004 |
| SLE | rs3747093 | rs7444 | 0.88 | UBE2L3 | rs5754102 | T | K4 | 22:22399916:22404237 |

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|-----|------------|------------|------|--------|-------------|---|-----|---|
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs7120313 | M | G | ENSG00000070047.6 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs2246614 | M | G | ENSG00000161328.10 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs11246177 | M | G | ENSG00000185507.14 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs936472 | M | G | ENSG00000185522.4 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs936472 | M | K27 | 11:600961:621989 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs386614207 | M | K4 | 11:601613:633623 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs2246614 | N | K27 | 11:600961:621989 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs11246217 | N | S | 11:614038:614173:2:2:1, 11:614038:614475:2:2:1 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs12290989 | N | S | 11:614038:614475:2:2:1, 11:614400:614475:2:2:1 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs4963128 | T | G | ENSG00000185507.14 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs12419618 | T | K27 | 11:600961:621989 |
| SLE | rs58688157 | rs12802200 | 0.48 | IRF7 | rs12803048 | T | K4 | 11:601613:633623 |
| SLE | rs6671847 | rs1801274 | 0.89 | FCGR2A | rs4657041 | M | S | 1:161512990:161595934:2:2:0, 1:161594430:161595934:2:2:1 |
| SLE | rs6671847 | rs1801274 | 0.89 | FCGR2A | rs12129787 | N | S | 1:161487928:161489591:1:1:0, 1:161488906:161489591:1:1:0, 1:161489451:161489591:1:1:1 |

Supplementary Table 2.2: Summary of all features colocalised with disease loci that colocalised with at least one gene or splicing QTL

All disease loci that colocalised with at least one gene or splicing QTL is summarised here. Loci colocalised with only histone features are not listed. The Disease SNP column describes the SNP assigned from the GWAS summary statistics (Materials and Methods). The Study SNP is the disease lead SNP listed in the study, and the LD is the 1000G LD between the Study SNP and Disease SNP if these differ. Differences may occur if the summary statistics available were a subset of a full study meta-analysis. The Study SNP was used to assign the locus, designated in the study. The Feature SNP is the molecular feature lead SNP. Cell is the cell-type of the corresponding feature (M = monocyte, N = neutrophil, T = T cell). Mark describes the feature type (G = Gene, S = splicing/PSI, K4 = H3K4me1, K27ac, H3K27ac). The feature ID describes the Ensembl gene ID or the histone signal peak defined as chr:start:end. The splicing ID describes the splicing junctions defined by Chen *et al* (2016a).

BLUEPRINT summary statistics for lead QTLs can be found here: <http://blueprint-dev.bioinfo.cnio.es/WP10/>

Lead study SNP summaries can be found here: AMD PMID: 26691988, AD PMID: 24162737, CAD PMID: 26343387, FEV₁ PMID: 26423011, SLE PMID: 26502338

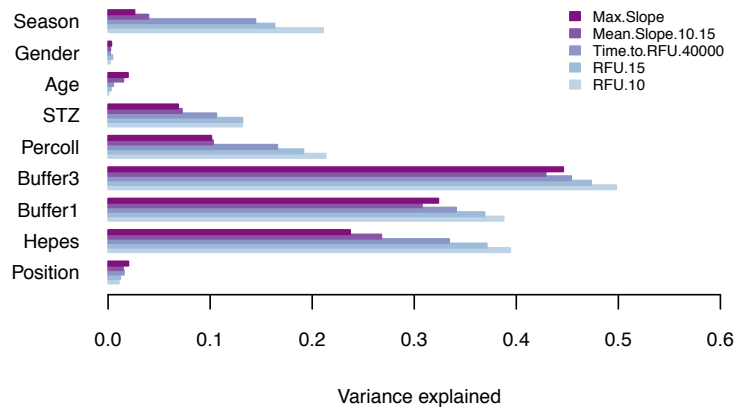
Freely downloadable Summary statistics: Alzheimer's Disease, Lambert *et al* (2013): http://web.pasteur-lille.fr/en/recherche/u744/igap/igap_download.php

Coronary artery disease, Nikpay *et al.* (2015): <http://www.cardiogramplusc4d.org/data-downloads/> Systemic lupus erythematosus, Bentham *et al.* (2015):

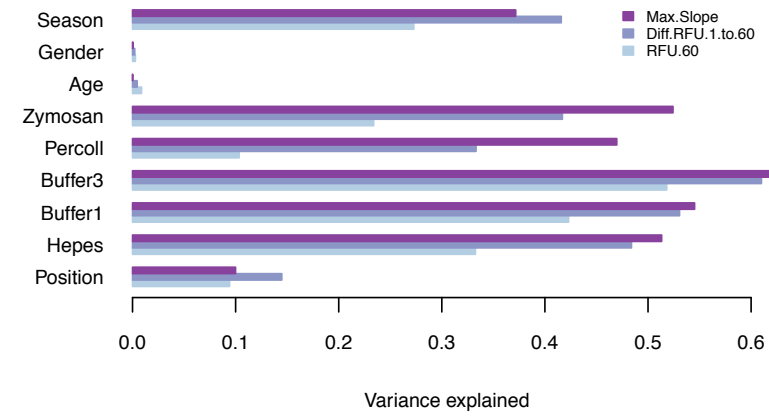
https://www.immunobase.org/downloads/protected_data/GWAS_Data/

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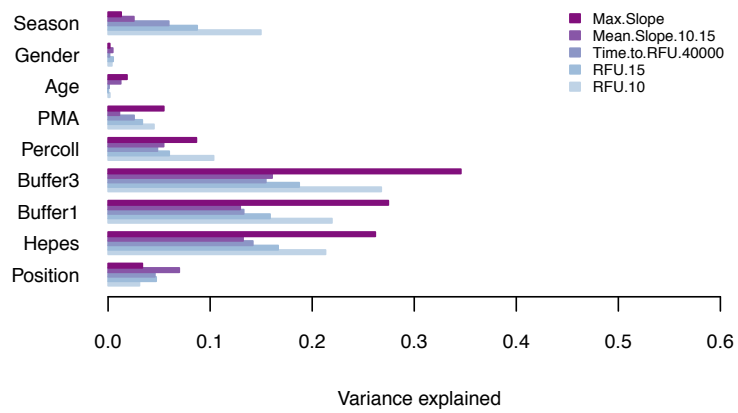
Respiratory burst STZ



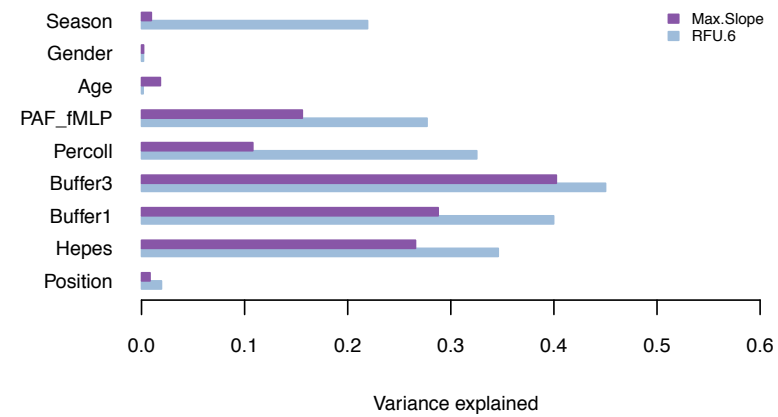
Respiratory burst Zymosan



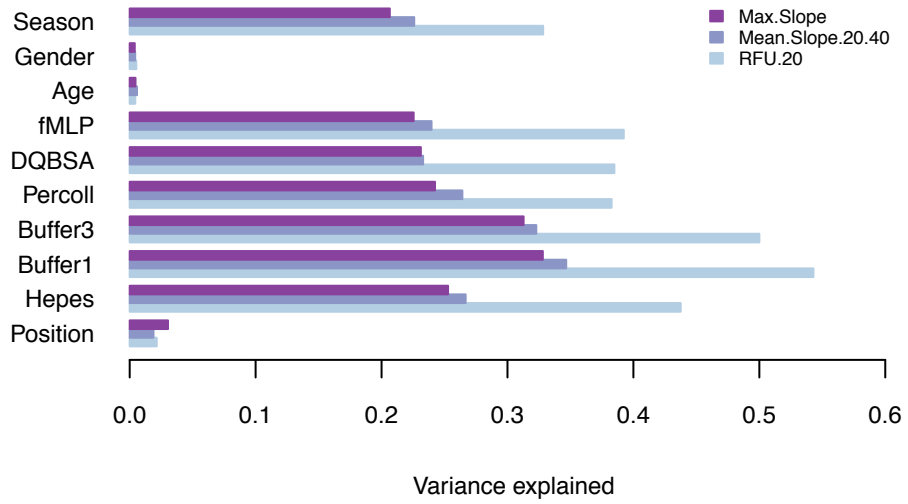
Respiratory burst PMA



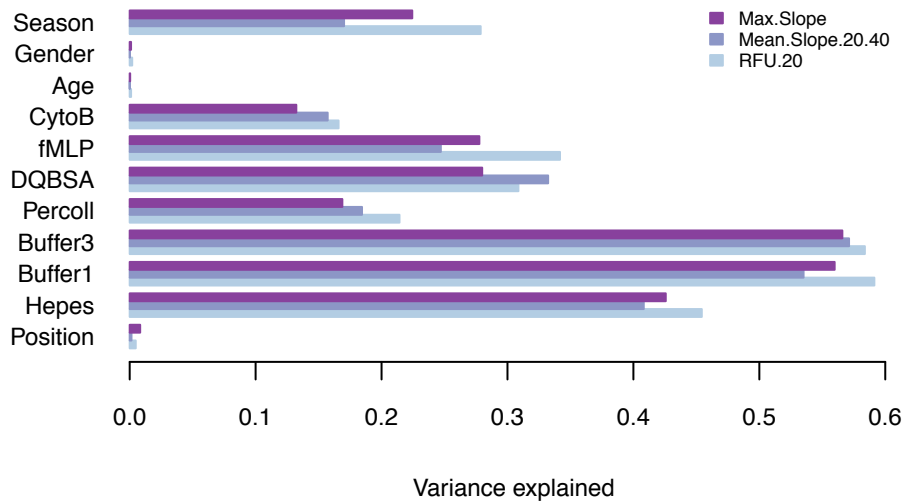
Respiratory burst PAF + fMLP



Degranulation fMLP

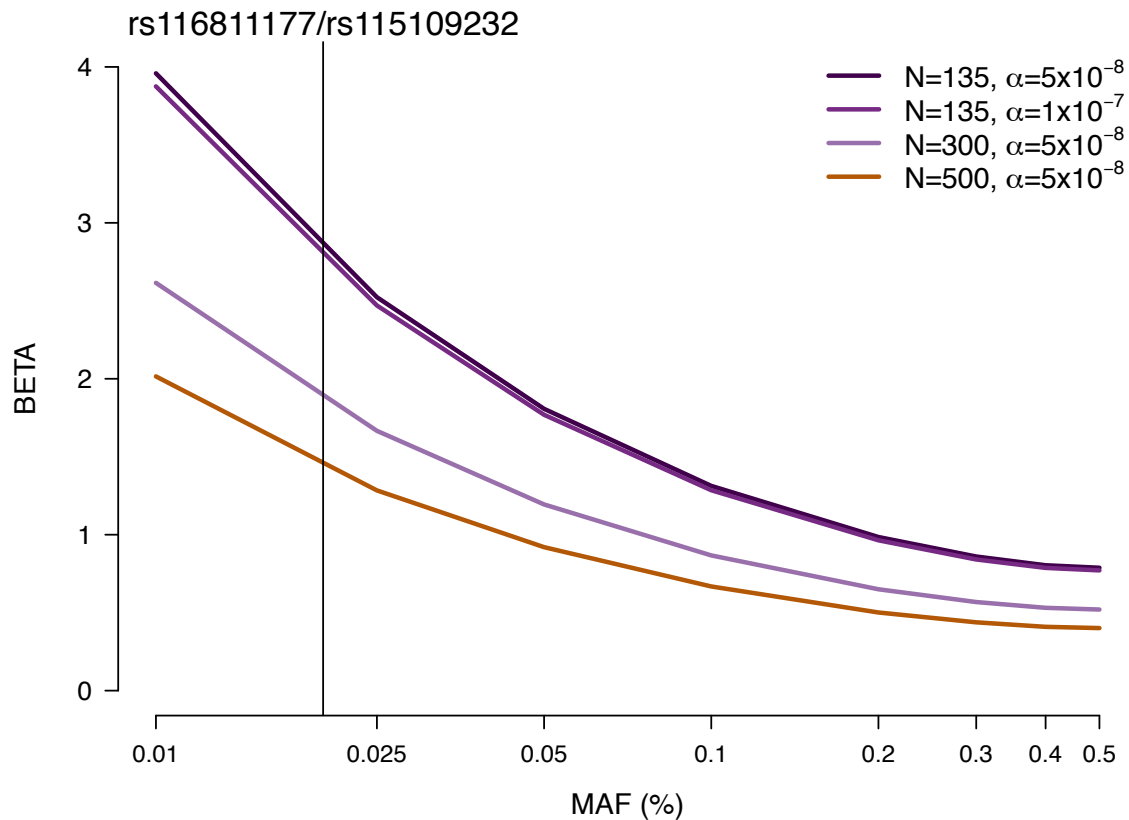


Degranulation CytoB + fMLP



Supplementary Figure 3.1: Contribution of known sources of co-variation to respiratory burst and degranulation neutrophil responses

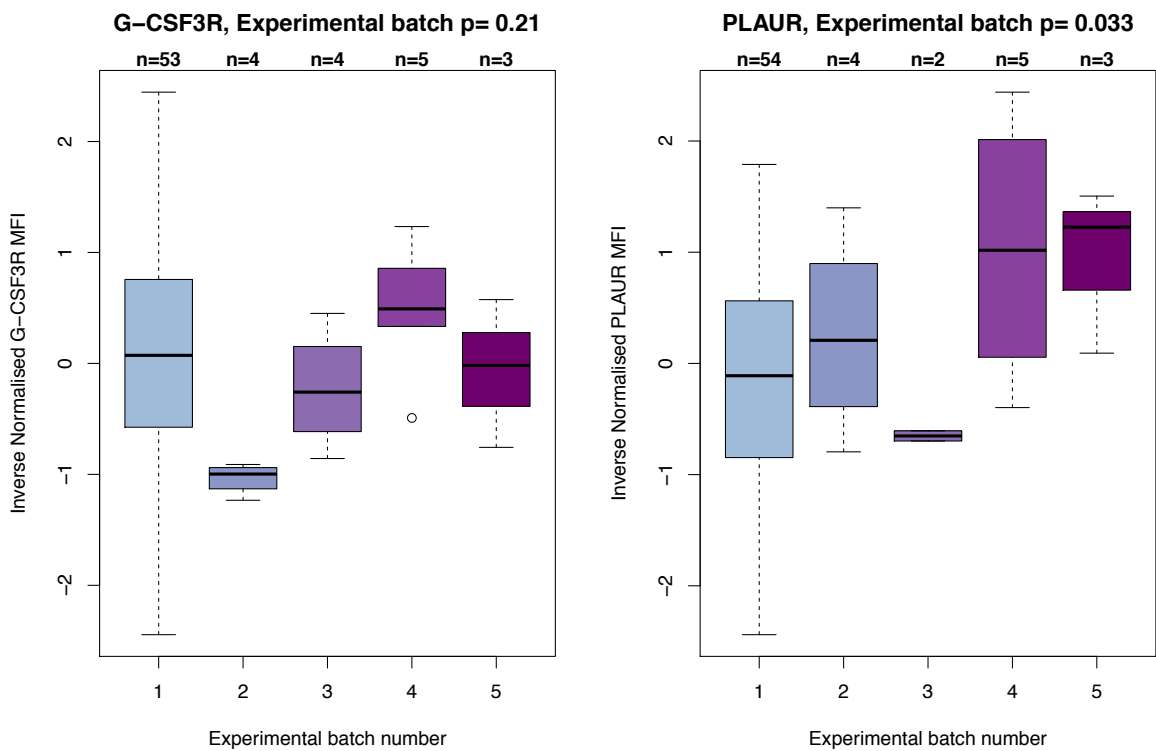
Barplots show R^2 values from fitting linear models of each trait with each covariate independently.



Supplementary Figure 3.2: Predicted power of the neutrophil function study

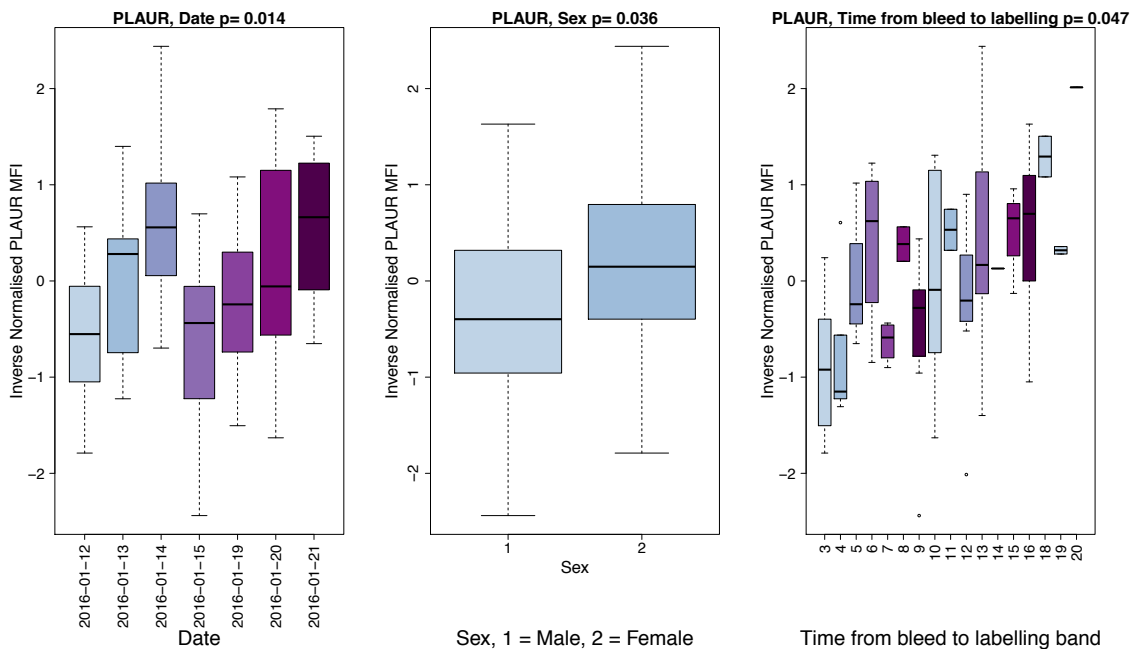
A power calculation was performed for a study of 135 individuals (approximately equal to this function cohort) and for increasing sample sizes up to 500 individuals. Variants will be likely detected by the study if the variant falls above the line which describes the relationship between beta (effect size in standard deviation of the trait value) and minor allele frequency. This study is powered to detect common variants for moderate effect size or low frequency variants with higher effect sizes of beta greater than 2. The power for the suggestive p value threshold of 1×10^{-07} for $N = 135$ is also shown. The corresponding frequency of the genome-wide significant locus (rs116811177/rs115109232) identified in this study is marked. The beta for the association for both SNPs from this study was 2.92, which is close to the predicted power of this study to detect SNPs of this low-frequency. The pwr package was used in this power calculation (Champely, 2012).

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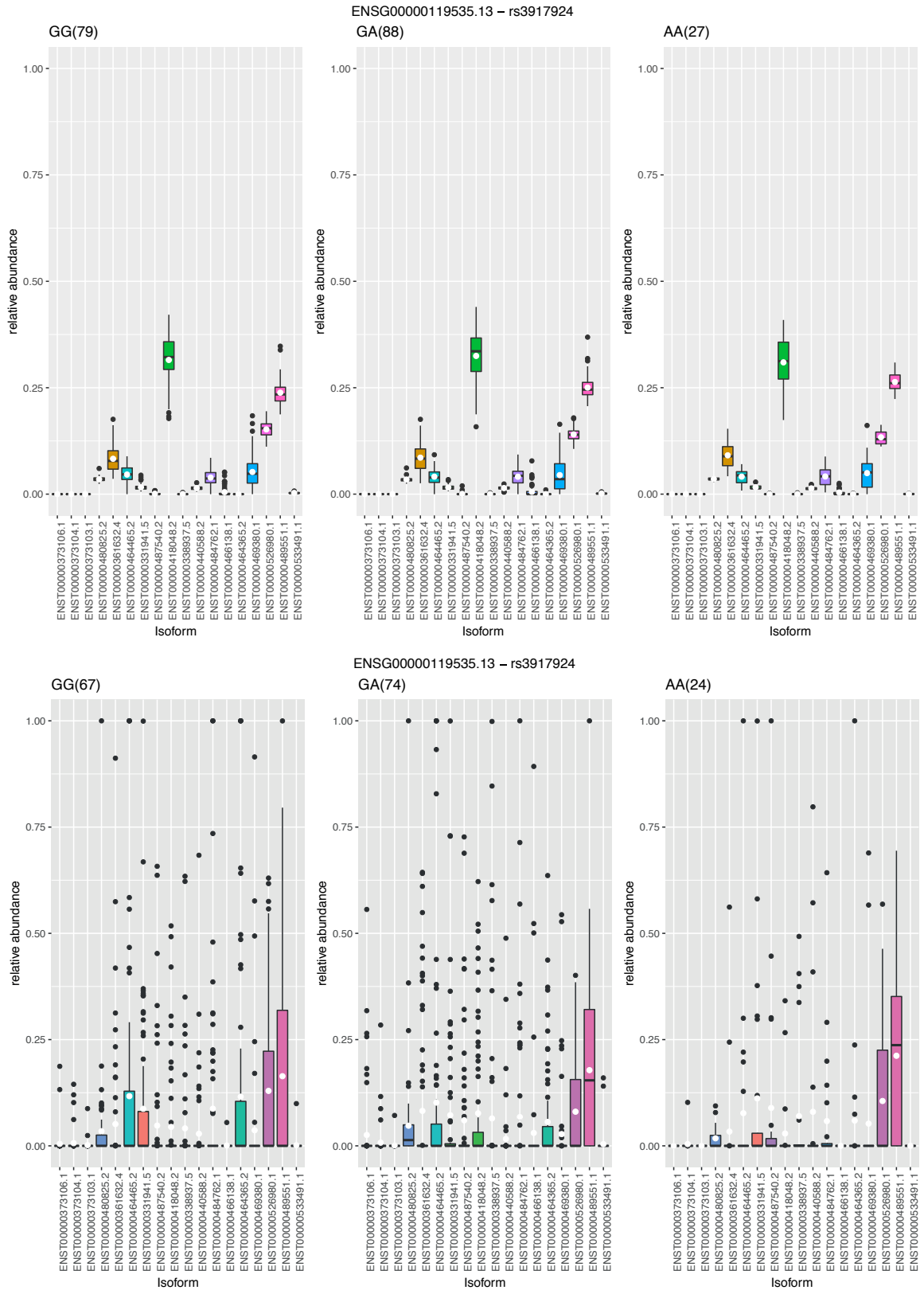
Supplementary Figure 4.1: Receptor MFI values stratified by experimental processing batch

Normalised GCSFR and PLAUR MFI stratified by experimental processing batch with outliers removed from both datasets. ANOVA testing with inverse normalised trait values found this covariate to be significant for PLAUR MFI only.



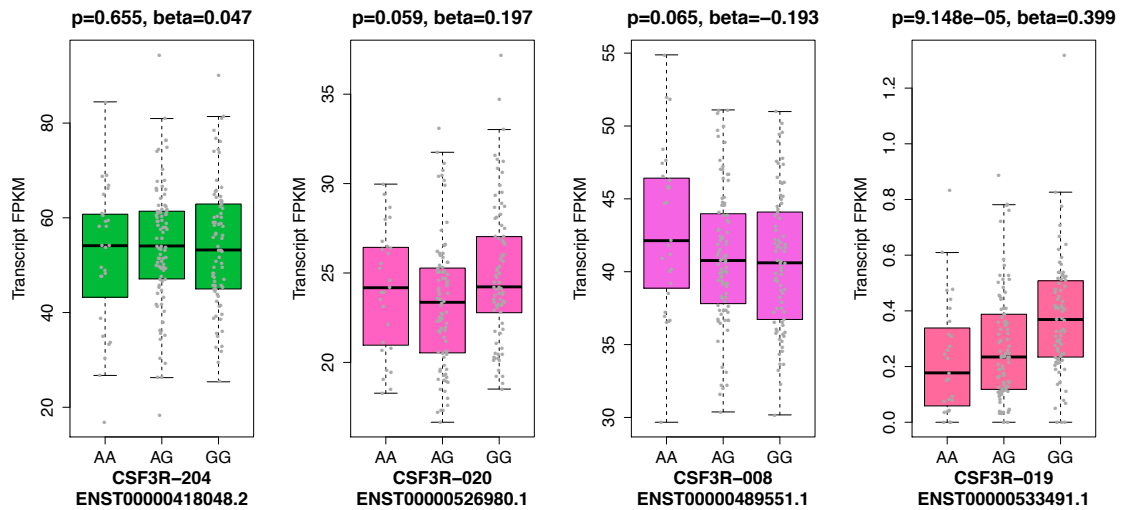
Supplementary Figure 4.2: Covariate effects on MFI values for PLAUR receptor

Normalised MFI values for the PLAUR receptor, excluding outliers, are shown stratified by date, sex and time from bleed to labelling, which were all found to be significant covariates using ANOVA. The p value for each covariate from the ANOVA testing is listed in the corresponding plots.



Supplementary Figure 4.3: Relative abundance of *CSF3R* transcripts in T cells and monocytes stratified by rs3917924 genotypes

This abundance by genotype plot shows all 18 *CSF3R* transcripts (Gencode v15) for T cells (top panel) and monocytes (bottom panel). rs3917924 is common GCSFR surface expression index SNP). The receptor-encoding transcript (green) is most abundantly expressed in monocytes but not highly expressed in T cells.



Supplementary Figure 4.4: CSF3R transcript expression level in monocytes

The three most abundant transcripts (left to right) including the short truncated transcript, CSF3R-019 (far right) are shown with the transcript expression level (FPKM) in monocytes stratified by the genotype of rs3917924. The p value and beta are shown after using a linear regression to test association of transcript level with genotype. Although the expression level of CSF-020 is significant ($\beta = 0.483$, $SE = 0.151$, p value 0.002), the effect does not look additive. CSF3R-019 transcript expression is significantly associated with rs3917924 genotype ($\beta = 0.48$, $SE = 0.147$, p value = 0.001) but the effect is much smaller than observed with neutrophils (Figure 4.12). The transcript expression levels of all four transcripts are lower than in neutrophils.

| SNP | Proteins bound | Motifs altered | Histone | CEBPB/PU1 | ChromHMM | HL60 TF | Differentiated HL60 |
|-----------|---------------------------------------|---|---|---|--|---|--|
| rs3917912 | | AP-2_disc2, ELF1_disc2, Ets_known3 | K27ac (1:36928496:36957321) K4me1 (1: 36778894:36993055) | PU1 (1: 36947299:36949245) | Active TSS high H3K4me3+K27Ac (1: 36947600:36948600) | P300 (1:36947664:36948535) CEBPE (1:36947667:36949236), H3K4me1, H3K4me3, H3K27ac | H3K27ac DMSO, H3K4me3 ATRA (1:36946936:36948821), H3K27ac ATRA, H3K4me1 DMSO, H3K4me3 ATRA |
| rs3917914 | | | K27ac (1:36928496:36957321) K4me1 (1: 36778894:36993055) | PU1 (1: 36947299:36949245) | Active TSS high H3K4me3+K27Ac (1: 36947600:36948600) | P300 (1:36947664:36948535) CEBPE (1:36947667:36949236), H3K4me1, H3K4me3, H3K27ac | H3K27ac DMSO, H3K4me3 ATRA (1:36946936:36948821), H3K27ac ATRA, H3K4me1 DMSO, H3K4me3 ATRA |
| rs3917924 | CEBPB, JUND, P300, TBP, BAF170 (Hela) | BCL_disc5, NRSF_disc3 | K27ac (1:36928496:36957321) K4me1 (1: 36778894:36993055) | PU1 (1: 36944990:36945853) CEBPB (1:36944999:36945946) | Active TSS High H3K4me3 (E10) (1:36945600:36945800) | CEBPE (1:36944824:36946074), H3K4me1, H3K4me3 | H3K4me1 ATRA, H3K4me1 DMSO |
| rs3917932 | | AP-1_disc1, ATF3_disc1, ATF3_known1, ATF3_known10, ATF3_known9, ATF4, ATF6, E2F_disc1, HEY1_disc1, LXR_2, Maf_disc2, Mxi1_known1, Myc_disc1, SIRT6_disc1, SREBP_known4, T3R, TATA_disc2 | K27ac (1:36928496:36957321) K4me1 (1: 36778894:36993055) | | Active Enh (K27ac, K4ME1,E9) (1:36943800:36944000) | H3K4me1 | |

Supplementary Table 4.1: Intersection of *CSF3R* significant variants with epigenomic and molecular data

Common and rare lead SNPs for GCSFR MFI and neutrophil count are listed and annotations/ First those using HaploReg v4.1 including predicted disrupted motifs (Keradpour and Kellis, 2014). Column four onwards are epigenome-SNP overlaps (bedtools intersect). The data used was for primary neutrophils from the BLUEPRINT consortium (Chen et al, 2016, Carrillo-de-Santa-Pau et al., 2017) as well as data from Stephen Watt (manuscript in preparation). The histone peaks used for intersection were those as tested for QTL in the blueprint consortium using a 1Mb window. The final two columns list epigenome-SNP overlaps of ChIP-seq data generated in the neutrophil model cell line, HL60 and the differentiated more-mature HL60 model (ATRA and DMSO) (Chapter 2).

| SNP | EA/OA | PU1 1Mb QTL | Exon | Splicing Junction | Allele-specific eQTL (WASP-ASE) |
|-----------|-------|--|---|--|---|
| rs3917924 | G/A | 1:36924227:36924948, 1.518 x 10 ⁻⁰⁶ , -0.6867 (peak falls 3' outside of CSF3R gene) | ENSG00000119535.13.46, 1.091 x 10 ⁻³⁷ , 1.104 ENSG00000119535.13.41, 4.353 x 10 ⁻⁰⁴ , 0.3637 | 1:36945118:36945587:2:2:1 4.487 x 10 ⁻⁵⁸ , 1.245 1:36945682:36947078:2:2:1 6.407 x 10 ⁻⁵⁵ , 1.231 1:36943279:36945033:2:2:1 2.056 x 10 ⁻⁰⁹ , 0.600 1:36941275:36943205:2:2:0 5.667 x 10 ⁻⁰⁶ , 0.463 | ENSG00000119535.13 5.587 x 10 ⁻³³ , 0.103 |
| rs3917931 | C/T | 1:36924227:36924948, 1.518 x 10 ⁻⁰⁶ , -0.6867 | ENSG00000119535.13.46, 1.483 x 10 ⁻³⁸ , 1.122 ENSG00000119535.13.41, 2.871 x 10 ⁻⁰⁴ , 0.378 | 1:36945118:36945587:2:2:1 1.687 x 10 ⁻⁵⁹ , 1.264 1:36945682:36947078:2:2:1 9.940 x 10 ⁻⁵⁶ , 1.247 1:36943279:36945033:2:2:1 7.018 x 10 ⁻¹⁰ , 0.622 1:36941275:36943205:2:2:0 4.664 x 10 ⁻⁰⁶ , 0.471 | ENSG00000119535.13 7.191 x 10 ⁻³² , 0.100 |
| rs3917925 | G/A | 1:36924227:36924948, 1.518 x 10 ⁻⁰⁶ , -0.6867 | ENSG00000119535.13.46, 1.483 x 10 ⁻³⁸ , 1.122 ENSG00000119535.13.41, 2.871 x 10 ⁻⁰⁴ , 0.378 | 1:36945118:36945587:2:2:1 1.687 x 10 ⁻⁵⁹ , 1.264 1:36945682:36947078:2:2:1 9.940 x 10 ⁻⁵⁶ , 1.247 1:36943279:36945033:2:2:1 7.018 x 10 ⁻¹⁰ , 0.622 1:36941275:36943205:2:2:0 4.664 x 10 ⁻⁰⁶ , 0.471 | ENSG00000119535.13 7.191 x 10 ⁻³² , 0.100 |
| rs6667127 | C/T | 1:36924227:36924948, 1.690 x 10 ⁻⁰⁵ , -0.6704 | ENSG00000119535.13.46, 3.067 x 10 ⁻²² , 0.969 | 1:36945118:36945587:2:2:1 1.407 x 10 ⁻³² , 1.119 1:36945682:36947078:2:2:1 2.881 x 10 ⁻³⁰ , 1.092 1:36943279:36945033:2:2:1 4.997 x 10 ⁻⁰⁶ , 0.500 1:36941275:36943205:2:2:0 1.349 x 10 ⁻⁰⁴ , 0.420 | ENSG00000119535.13 1.095 x 10 ⁻²² , 0.090 |
| rs955115 | C/A | 1:36924227:36924948, 5.247 x 10 ⁻⁰⁵ , -0.6151 | ENSG00000119535.13.46, 7.116 x 10 ⁻⁰⁸ , 0.5853 | 1:36945682:36947078:2:2:1 1.519 x 10 ⁻¹⁰ , 0.683 1:36945118:36945587:2:2:1 6.471 x 10 ⁻⁰⁹ , 0.623 1:36935442:36937033:2:2:1 1.899 x 10 ⁻⁰⁴ , 0.415 1:36943279:36945033:2:2:1 7.702 x 10 ⁻⁰⁴ , 0.374 | ENSG00000119535.13 3.852 x 10 ⁻⁰⁹ , 0.068 |
| rs3917933 | G/A | 1:36924227:36924948, 1.518 x 10 ⁻⁰⁶ , -0.6867 | ENSG00000119535.13.46, 1.483 x 10 ⁻³⁸ , 1.122 ENSG00000119535.13.41, 2.871 x 10 ⁻⁰⁴ , 0.378 | 1:36945118:36945587:2:2:1 1.687 x 10 ⁻⁵⁹ , 1.264 1:36945682:36947078:2:2:1 9.940 x 10 ⁻⁵⁶ , 1.247 1:36943279:36945033:2:2:1 7.018 x 10 ⁻¹⁰ , 0.622 1:36941275:36943205:2:2:0 4.664 x 10 ⁻⁰⁶ , 0.471 | ENSG00000119535.13 7.191 x 10 ⁻³² , 0.100 |

Supplementary Table 4.2: Summary of significant QTL associations within the GCSFR locus that were tested in the BLUEPRINT consortium

Significant QTL associations with the common SNPs from the BLUEPRINT consortium (Chen et al. 2016). Features with a qvalue of 5% or less are listed. For comparison, the uncorrected raw p value of association is listed for each feature. The phenotype for PU1 is the binding of this transcription factor assayed using ChIP-seq in primary neutrophils in a subset of the same individuals as the main Blueprint consortium (Stephen Watt, manuscript in preparation). 1Mb refers to the window size around the feature for the QTLs tested. Exon and splicing junction QTLs were identified using RNA-seq data and processed in the same way as gene expression QTLs from Chen *et al.* (2016), but the reads for exon QTLs are summed over each individual exon in a gene and over each splicing junction for the latter QTLs. These were not published as part of the Chen *et al.* (2016) paper. Allele-specific eQTLs were published as part of the main study and analysed using the WASP software.

